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6 FILES IN THE FILE LIST

=> s PXE or pseudoxanthoma or pseudoxanthoma eleasticum
L1        3778 PXE OR PSEUDOXANTHOMA OR PSEUDOXANTHOMA ELEASTICUM

=> s l1 and (MRP6 or ABCC6)
L2         487 L1 AND (MRP6 OR ABCC6)

=> s l2 and R1141X
L3          68 L2 AND R1141X

=> dup rem l3
PROCESSING COMPLETED FOR L3
L4          22 DUP REM L3 (46 DUPLICATES REMOVED)

=> d ibib abs l4 1-22

L4   ANSWER 1 OF 22     MEDLINE on STN
ACCESSION NUMBER:    2006495014     MEDLINE
DOCUMENT NUMBER:     PubMed ID: 16835894
TITLE:               Mutational analysis of the ABCC6 gene and the
                     proximal ABCC6 gene promoter in German patients
                     with pseudoxanthoma elasticum (PXE).
AUTHOR:              Schulz Veronika; Hendig Doris; Henjakovic Maja; Szliska
                     Christiane; Kleesiek Knut; Gotting Christian
CORPORATE SOURCE:    Institut fur Laboratoriums- und Transfusionsmedizin, Herz-
                     und Diabeteszentrum Nordrhein-Westfalen, Universitatsklinik
                     der Ruhr-Universitat Bochum, Bad Oeynhausen, Germany.
SOURCE:              Human mutation, (2006 Aug) Vol. 27, No. 8, pp. 831.
                     Journal code: 9215429. E-ISSN: 1098-1004.
PUB. COUNTRY:        United States
DOCUMENT TYPE:       Journal; Article; (JOURNAL ARTICLE)
LANGUAGE:            English
FILE SEGMENT:        Priority Journals
ENTRY MONTH:         200610
ENTRY DATE:          Entered STN: 22 Aug 2006
                     Last Updated on STN: 17 Oct 2006
                     Entered Medline: 16 Oct 2006
AB   Pseudoxanthoma elasticum (PXE) is a genetic disorder
     characterized by calcification of elastic fibers in dermal, ocular, and
     cardiovascular tissues.  Recently, ABCC6 mutations were
     identified as causing PXE.  In this follow-up study we report
     the investigation of 61 German PXE patients from 53 families,
     hitherto the largest cohort of German PXE patients screened for
     the complete ABCC6 gene.  In addition, we characterized the
     proximal ABCC6 promoter of PXE patients according to
     mutation.  In this study we identified 32 disease-causing ABCC6
     variants, which had been described previously by us and others, and 10
     novel mutations (eight missense mutations and two splice site
     alterations).  The mutation detection rate among index patients was 87.7%.
     Frequent alterations were the PXE-mutations p.R1141X,
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NEWS 8	NOV 20	CAS Registry Number crossover limit increased to 300,000 in additional databases
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NEWS 11	DEC 11	CAS REGISTRY chemical nomenclature enhanced
NEWS 12	DEC 14	WPIDS/WPINDEX/WPIX manual codes updated
NEWS 13	DEC 14	GBFULL and FRFULL enhanced with IPC 8 features and functionality
NEWS 14	DEC 18	CA/CAplus pre-1967 chemical substance index entries enhanced with preparation role
NEWS 15	DEC 18	CA/CAplus patent kind codes updated
NEWS 16	DEC 18	MARPAT to CA/CAplus accession number crossover limit increased to 50,000
NEWS 17	DEC 18	MEDLINE updated in preparation for 2007 reload
NEWS 18	DEC 27	CA/CAplus enhanced with more pre-1907 records
NEWS 19	JAN 08	CHEMLIST enhanced with New Zealand Inventory of Chemicals
NEWS 20	JAN 16	CA/CAplus Company Name Thesaurus enhanced and reloaded
NEWS 21	JAN 16	IPC version 2007.01 thesaurus available on STN
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NEWS HOURS	STN Operating Hours Plus Help Desk Availability
NEWS LOGIN	Welcome Banner and News Items
NEWS IPC8	For general information regarding STN implementation of IPC 8
NEWS X25	X.25 communication option no longer available

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Ex23_Ex29del, and c.2787+1G > T. In the ABCC6 promoter we found the polymorphisms c.-127C > T, c.-132C > T, and c.-219A > C. The difference in the c.-219A > C frequencies between PXE patients and controls were determined as statistically significant. Interestingly, c.-219A > C is located in a transcriptional activator sequence of the ABCC6 promoter and occurred in a binding site for a transcriptional repressor, predominantly found in genes that participate in lipid metabolism. Obtaining these genetic data signifies our contribution to elucidating the pathogenetics of PXE.

L4 ANSWER 2 OF 22 MEDLINE on STN DUPLICATE 1
ACCESSION NUMBER: 2006064186 MEDLINE
DOCUMENT NUMBER: PubMed ID: 16384891
TITLE: Role of serum fetuin-A, a major inhibitor of systemic calcification, in pseudoxanthoma elasticum.
AUTHOR: Hendig Doris; Schulz Veronika; Arndt Marius; Szliska Christiane; Kleesiek Knut; Gotting Christian
CORPORATE SOURCE: Institut fur Laboratoriums- und Transfusionsmedizin, Herz- und Diabeteszentrum Nordrhein-Westfalen, Universitatsklinik der Ruhr-Universitat Bochum, Bad Oeynhausen, Germany.
SOURCE: Clinical chemistry, (2006 Feb) Vol. 52, No. 2, pp. 227-34. Electronic Publication: 2005-12-29. Journal code: 9421549. ISSN: 0009-9147.
PUB. COUNTRY: United States
DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE)
LANGUAGE: English
FILE SEGMENT: Priority Journals
ENTRY MONTH: 200603
ENTRY DATE: Entered STN: 2 Feb 2006
Last Updated on STN: 11 Mar 2006
Entered Medline: 10 Mar 2006

AB BACKGROUND: Pseudoxanthoma elasticum (PXE) is a hereditary disorder of the connective tissue affecting the skin, retina, and cardiovascular system and characterized by progressive calcification of abnormal and fragmented elastic fibers in the extracellular matrix. The aim of the present study was to investigate the association of fetuin-A, a major systemic inhibitor of calcification, with PXE. METHODS: Fetuin-A was measured by quantitative sandwich enzyme immunoassay in sera from 110 German patients with PXE, 53 unaffected first-degree family members, and 80 healthy blood donors. We determined the distribution of the fetuin-A polymorphisms c.742C>T (p.T248M) and c.766C>G (p.T256S) in these same 3 groups. The occurrences of the frequent ABCC6 gene mutations c.3421C>T (p.R1141X) and c.EX23_EX29del were also assessed. RESULTS: Serum fetuin-A concentrations in male and female PXE patients were lower than in unaffected first-degree relatives and controls [mean (SD) concentrations, 0.55 (0.11) g/L in patients; 0.70 (0.23) g/L in relatives; and 0.80 (0.23) g/L in controls (P <0.0001)]. Serum fetuin-A was higher in female PXE patients with cardiovascular involvement than in the corresponding male group (P <0.05). The fetuin-A polymorphism frequencies did not differ among PXE patients, family members, and blood donors. CONCLUSION: A deficiency of multidrug resistance-associated protein 6 leads to alteration of circulating substrates, e.g., inhibitors of calcification as fetuin-A, leading to progressive mineralization of elastic fibers in PXE.

L4 ANSWER 3 OF 22 MEDLINE on STN DUPLICATE 2
ACCESSION NUMBER: 2005641044 MEDLINE
DOCUMENT NUMBER: PubMed ID: 16133423
TITLE: Elevated xylosyltransferase I activities in pseudoxanthoma elasticum (PXE) patients as a marker of stimulated proteoglycan biosynthesis.
AUTHOR: Gotting Christian; Hendig Doris; Adam Alexandra; Schon Sylvia; Schulz Veronika; Szliska Christiane; Kuhn Joachim;

Kleesiek Knut
CORPORATE SOURCE: Institut fur Laboratoriums-und Transfusionsmedizin, Herz-und Diabeteszentrum Nordrhein-Westfalen, Universitatsklinik der Ruhr-Universitat Bochum, Georgstrasse 11, 32545 Bad Oeynhausen, Germany.. cgoetting@hdz-nrw.de
SOURCE: Journal of molecular medicine (Berlin, Germany), (2005 Dec) Vol. 83, No. 12, pp. 984-92. Electronic Publication: 2005-08-24.
Journal code: 9504370. ISSN: 0946-2716.
PUB. COUNTRY: Germany: Germany, Federal Republic of
DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE)
LANGUAGE: English
FILE SEGMENT: Priority Journals
ENTRY MONTH: 200605
ENTRY DATE: Entered STN: 3 Dec 2005
Last Updated on STN: 4 May 2006
Entered Medline: 3 May 2006

AB Pseudoxanthoma elasticum (PXE) is a hereditary disorder of the connective tissue characterized by extracellular matrix alterations with elastin fragmentation and excessive proteoglycan deposition. Xylosyltransferase I (XT-I, E.C. 2.4.2.26) is the initial enzyme in the biosynthesis of the glycosaminoglycan chains in proteoglycans and has been shown to be a marker of tissue remodeling processes. Here, we investigated for the first time serum XT-I activities in a large cohort of German PXE patients and their unaffected relatives. XT-I activities were measured in serum samples from 113 Caucasian patients with PXE and 103 unaffected first-degree family members. The occurrence of the frequent ABCC6 gene mutation c.3421C>T (R1141X) and the hypertension-associated genetic variants T174M and M235T in the angiotensinogen (AGT) gene were determined. Serum XT-I activities in male and female PXE patients were significantly increased compared to unaffected family members (male patients, mean value 0.96 mU/l, SD 0.37; male relatives, 0.78 mU/l, SD 0.29; female patients, 0.91 mU/l, SD 0.31; female relatives, 0.76 mU/l, SD 0.34; $p<0.05$). The mean XT-I activities in PXE patients with hypertension were 24% higher than in patients without increased blood pressure ($p<0.05$). The AGT T174M and M235T frequencies were not different in hypertensive PXE patients, normotensive PXE patients, family members or blood donors. Our data show that the altered proteoglycan biosynthesis in PXE patients is closely related to an increased XT-I activity in blood. Serum XT-I, the novel fibrosis marker, may be useful for the assessment of extracellular matrix alterations and disease activity in PXE.

L4 ANSWER 4 OF 22 MEDLINE on STN DUPLICATE 3
ACCESSION NUMBER: 2005202853 MEDLINE
DOCUMENT NUMBER: PubMed ID: 15837081
TITLE: Patients with premature coronary artery disease who carry the ABCC6 R1141X mutation have no Pseudoxanthoma Elasticum phenotype.
AUTHOR: Wegman Jurgen J; Hu Xiaofeng; Tan Hendra; Bergen Arthur A B; Trip Mieke D; Kastelein John J P; Smulders Yvo M
CORPORATE SOURCE: Department of Vascular Medicine, Academic Medical Center, University of Amsterdam, The Netherlands.
SOURCE: International journal of cardiology, (2005 Apr 28) Vol. 100, No. 3, pp. 389-93.
Journal code: 8200291. ISSN: 0167-5273.
PUB. COUNTRY: Ireland
DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE)
LANGUAGE: English
FILE SEGMENT: Priority Journals
ENTRY MONTH: 200507
ENTRY DATE: Entered STN: 20 Apr 2005

Last Updated on STN: 29 Jul 2005
Entered Medline: 28 Jul 2005

AB BACKGROUND: Pseudoxanthoma elasticum (PXE) is an inherited disorder of elastic tissue. We recently found that heterozygosity for the frequent (0.8% prevalence in Dutch population) R1141X mutation in the PXE gene coding for the ABCC6 transporter, is associated with a fourfold risk of premature coronary artery disease. Yet, it is not clear whether or not heterozygosity for this mutation results in a mild PXE phenotype. The objective of our study was to determine if skin and/or eye abnormalities related to a PXE phenotype could be found in patients with premature coronary artery disease, with and without the R1141X mutation. METHODS: R1141X mutation carriers with premature coronary artery disease (cases) and patients with premature coronary artery disease with no-or not known-mutation (controls) were studied. Cases and controls were examined for PXE-like skin changes and retinal angioid streaks, peau d'orange or pigment epithelium changes. RESULTS: 7 cases and 31 controls were analysed. In both the mutation-positive and the control group, skin inspection and eye fundus examination did not reveal any dermatological or ocular signs of PXE. CONCLUSIONS: Carriers for the ABCC6 R1141X mutation, which is frequent and confers a high risk of premature coronary artery disease, do not commonly have skin or eye abnormalities consistent with a mild PXE phenotype.

L4 ANSWER 5 OF 22 MEDLINE on STN DUPLICATE 4

ACCESSION NUMBER: 2006006847 MEDLINE
DOCUMENT NUMBER: PubMed ID: 16392638
TITLE: Novel mutations in the ABCC6 gene of German patients with pseudoxanthoma elasticum.
AUTHOR: Schulz Veronika; Hendig Doris; Szliska Christiane; Gotting Christian; Kleesiek Knut
CORPORATE SOURCE: Institut fur Laboratoriums- und Transfusionsmedizin, Herz- und Diabeteszentrum Nordrhein-Westfalen, Universitatsklinik der Ruhr-Universitat Bochum, Georgstrasse 11, 32545 Bad Oeynhausen, Germany.
SOURCE: Human biology; an international record of research, (2005 Jun) Vol. 77, No. 3, pp. 367-84.
Journal code: 0116717. ISSN: 0018-7143.
PUB. COUNTRY: United States
DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE)
LANGUAGE: English
FILE SEGMENT: Priority Journals
ENTRY MONTH: 200602
ENTRY DATE: Entered STN: 6 Jan 2006
Last Updated on STN: 22 Feb 2006
Entered Medline: 21 Feb 2006

AB Pseudoxanthoma elasticum (PXE) is a heritable disorder of the connective tissue affecting the skin, eyes, and cardiovascular system. Recently, the PXE candidate gene ABCC6 was identified and a limited number of ABCC6 mutations were observed in different PXE cohorts. To identify novel PXE -causing ABCC6 mutations in German patients with PXE, we investigated a cohort of 54 German PXE patients and 23 family members from 49 apparently nonconsanguineous families. From the mutational analysis we found 27 different ABCC6 sequence variations. Among these, 11 were polymorphisms or neutral alterations and 16 were PXE-causing mutations. The most common mutation in our PXE cohort was the nonsense mutation p.R1141X, which occurred with an allele frequency of 25.9%. Furthermore, we found nine missense, one additional nonsense, and two putative splice site mutations as well as three single-nucleotide deletions. Most of these mutations were unique and occurred in cytoplasmic regions of the MRP6 protein; these mutations are proposed to be critical for the physiological

function of the MRP6 protein. In these regions we also found the three novel PXE-causing mutations p.R1114C, p.Y1239H, and p.G1311E, which were identified in three alleles from patients with PXE and were absent in 200 healthy control subjects. In addition, the first genotype-phenotype correlation was observed. By obtaining these genetic mutation data, we are contributing to an overview of all ABCC6 mutations leading to PXE and the pathogenetics of this disease.

L4 ANSWER 6 OF 22 CAPLUS COPYRIGHT 2007 ACS on STN
ACCESSION NUMBER: 2005:1107490 CAPLUS
DOCUMENT NUMBER: 144:126802
TITLE: Molecular genetics of pseudoxanthoma elasticum: type and frequency of mutations in ABCC6
AUTHOR(S): Miksch, Sara; Lumsden, Amanda; Guenther, Ulf P.; Foernzler, Dorothee; Christen-Zaech, Stephanie; Daugherty, Carol; Ramesar, Rajkumar S.; Lebwohl, Mark; Hohl, Daniel; Neldner, Kenneth H.; Lindpaintner, Klaus; Richards, Robert I.; Struk, Berthold
CORPORATE SOURCE: Charite, Franz Volhard Clinic, HELIOS Klinikum, Humboldt University Berlin, Germany
SOURCE: Human Mutation (2005), 26(3), 235-248
CODEN: HUMUE3; ISSN: 1059-7794
PUBLISHER: Wiley-Liss, Inc.
DOCUMENT TYPE: Journal
LANGUAGE: English
AB Pseudoxanthoma elasticum (PXE) is a systemic heritable disorder that affects the elastic tissue in the skin, eye, and cardiovascular system. Mutations in the ABCC6 gene cause PXE. We performed a mutation screen in ABCC6 using haplotype anal. in conjunction with direct sequencing to achieve a mutation detection rate of 97%. This screen consisted of 170 PXE chromosomes in 81 families, and detected 59 distinct mutations (32 missense, eight nonsense, and six likely splice-site point mutations; one small insertion; and seven small and five large deletions). Forty-three of these mutations are novel variants, which increases the total number of PXE mutations to 121. While most mutations are rare, three nonsense mutations, a splice donor site mutation, and the large deletion comprising exons 23-29 (c.2996_4208del) were identified as relatively frequent PXE mutations at 26%, 5%, 3.5%, 3%, and 11%, resp. Chromosomal haplotyping with two proximal and two distal polymorphic markers flanking ABCC6 demonstrated that most chromosomes that carry these relatively frequent PXE mutations have related haplotypes specific for these mutations, which suggests that these chromosomes originate from single founder mutations. The types of mutations found support loss-of-function as the mol. mechanism for the PXE phenotype. In 76 of the 81 families, the affected individuals were either homozygous for the same mutation or compound heterozygous for two mutations. In the remaining five families with one uncovered mutation, affecteds showed allelic compound heterozygosity for the cosegregating PXE haplotype. This demonstrates pseudo-dominance as the relevant inheritance mechanism, since disease transmission to the next generation always requires one mutant allelic variant from each parent. In contrast to other previous clin. and mol. claims, our results show evidence only for recessive PXE. This has profound consequences for the genetic counseling of families with PXE.
REFERENCE COUNT: 46 THERE ARE 46 CITED REFERENCES AVAILABLE FOR THIS RECORD. ALL CITATIONS AVAILABLE IN THE RE FORMAT

L4 ANSWER 7 OF 22 MEDLINE on STN DUPLICATE 5
ACCESSION NUMBER: 2004471837 MEDLINE
DOCUMENT NUMBER: PubMed ID: 15382558
TITLE: [From gene to disease; pseudoxanthoma elasticum

and the ABCC6 gene].
Van gen naar ziekte; pseudoxanthoma elasticum en het ABCC6-gen.

AUTHOR: Bergen A A B; Plomp A S; Gorgels T G M F; de Jong P T V M
CORPORATE SOURCE: Interuniversitair Oogheelkundig Instituut, Meibergdreef 47, 11o05 BA Amsterdam.. a.bergen@ioi.knaw.nl
SOURCE: Nederlands tijdschrift voor geneeskunde, (2004 Aug 7) Vol. 148, No. 32, pp. 1586-9. Ref: 3
Journal code: 0400770. ISSN: 0028-2162.
PUB. COUNTRY: Netherlands
DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE)
General Review; (REVIEW)
LANGUAGE: Dutch
FILE SEGMENT: Priority Journals
ENTRY MONTH: 200410
ENTRY DATE: Entered STN: 23 Sep 2004
Last Updated on STN: 13 Oct 2004
Entered Medline: 12 Oct 2004

AB Pseudoxanthoma elasticum (PXE) is a hereditary disease of the connective tissue characterized by progressive dystrophic mineralization of elastic fibres. PXE patients have skin lesions, may experience loss of visual acuity and cardiovascular complications. The inheritance pattern of PXE is almost always autosomal recessive. In less than 2% of the families, PXE may be inherited in an autosomal dominant fashion. PXE is caused by mutations in the ABCC6 (MRP6) gene. The R1141X mutation is by far the most common mutation; it has been identified in 19 patients, or 30% of all PXE-patients in the Netherlands. The molecular pathology of PXE is complicated by yet unknown factors causing a variable clinical expression of the disease. In 80% of the 110 PXE patients the authors studied, at least one ABCC6 mutation was found. Molecular diagnostics of PXE is especially useful to confirm the clinical diagnosis.

L4 ANSWER 8 OF 22 CAPLUS COPYRIGHT 2007 ACS on STN
ACCESSION NUMBER: 2005:90130 CAPLUS
DOCUMENT NUMBER: 143:57937
TITLE: ABCC6 mutations in Italian families affected by pseudoxanthoma elasticum (PXE)
AUTHOR(S): Gheduzzi, Dealba; Giudetti, Rita; Anzivino, Claudia; Tarugi, Patrizia; di Leo, Enza; Quaglino, Daniela; Ronchetti, Ivonne Pasquali
CORPORATE SOURCE: Dept. Biomedical Sciences, University of Modena and Reggio Emilia, Modena, Italy
SOURCE: Human Mutation (2004), 24(5), 755/1-755/10
CODEN: HUMUE3; ISSN: 1059-7794
PUBLISHER: Wiley-Liss, Inc.
DOCUMENT TYPE: Journal
LANGUAGE: English

AB Pseudoxanthoma elasticum (PXE) is a genetic disorder, characterized by cutaneous, ocular and cardiovascular clin. symptoms, caused by mutations in a gene (ABCC6) that encodes for MRP6 (Multidrug Resistance associated Protein 6), an ATP-binding cassette membrane transporter. The ABCC6 gene was sequenced in 38 unrelated PXE Italian families. The mutation detection rate was 82.9%. Mutant alleles occurred in homozygous, compound heterozygous and heterozygous forms, however the great majority of patients were compound heterozygotes. Twenty-three different mutations were identified, among which 11 were new. Fourteen were missense (61%); five were nonsense (22%); two were frameshift (8.5%) and two were putative splice site mutations (8.5%). The great majority of mutations were located from exon 24 to 30, exon 24 being the most affected. Among the others, exons 9 and 12 were particularly involved. Almost all mutations were located in the intracellular site of MRP6. A pos. correlation was observed

between patient's age and severity of the disorder, especially for eye alterations. The relevant heterogeneity in clin. manifestations between patients with identical ABCC6 mutations, even within the same family, seems to indicate that, apart from PXE causative mutations, other genes and/or metabolic pathways might influence the clin. expression of the disorder.

REFERENCE COUNT: 31 THERE ARE 31 CITED REFERENCES AVAILABLE FOR THIS RECORD. ALL CITATIONS AVAILABLE IN THE RE FORMAT

L4 ANSWER 9 OF 22 MEDLINE on STN DUPLICATE 6

ACCESSION NUMBER: 2004191545 MEDLINE
DOCUMENT NUMBER: PubMed ID: 15086542
TITLE: Novel ABCC6 mutations in pseudoxanthoma elasticum.
AUTHOR: Chassaing Nicolas; Martin Ludovic; Mazereeuw Juliette; Barrie Laurence; Nizard Sonia; Bonafe Jean-Louis; Calvas Patrick; Hovnanian Alain
CORPORATE SOURCE: Department of Medical Genetics, Purpan Hospital, Toulouse, France.
SOURCE: The Journal of investigative dermatology, (2004 Mar) Vol. 122, No. 3, pp. 608-13.
Journal code: 0426720. ISSN: 0022-202X.
PUB. COUNTRY: United States
DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE)
LANGUAGE: English
FILE SEGMENT: Priority Journals
ENTRY MONTH: 200405
ENTRY DATE: Entered STN: 17 Apr 2004
Last Updated on STN: 26 May 2004
Entered Medline: 25 May 2004

AB Pseudoxanthoma elasticum (PXE) is a heritable connective tissue disorder caused by mutations in an ABC (ATP-Binding Cassette) transporter gene (ABCC6), which manifests with cutaneous, ophthalmologic, and cardiovascular findings. We studied a cohort of 19 families with PXE, and identified 16 different mutations, nine of which were novel variants. The mutation detection rate was about 77%. We found that arginine codon 518 was, with the previously described R1141X and EX23_29del, a recurrently mutated amino acid (11.5% of the mutations detected for each variant R518Q and R518X). No clear delineation of genotype/phenotype correlation was identified, and marked intra-familial variability of the disease was seen in one family. One family with pseudodominant inheritance displayed three distinct ABCC6 mutations, providing further evidence for the probable exclusive recessive transmission of PXE. These data contribute to the expanding database of ABCC6 mutations, to the description of phenotypic variability, and inheritance in PXE, and should be helpful for genetic counselling.

L4 ANSWER 10 OF 22 SCISEARCH COPYRIGHT (c) 2007 The Thomson Corporation on STN

ACCESSION NUMBER: 2004:409583 SCISEARCH
THE GENUINE ARTICLE: 816IH
TITLE: Does autosomal dominant pseudoxanthoma elasticum exist?
AUTHOR: Plomp A S; Hu X F; de Jong P T V M (Reprint); Bergen A A B
CORPORATE SOURCE: Netherlands Ophthalm Res Inst, KNAW, Meibergdreef 47, NL-1105 BA Amsterdam, Netherlands (Reprint); Netherlands Ophthalm Res Inst, KNAW, NL-1105 BA Amsterdam, Netherlands; Univ Amsterdam, Acad Med Ctr, Dept Clin Genet, NL-1105 AZ Amsterdam, Netherlands; Univ Amsterdam, Acad Med Ctr, Dept Ophthalmol, NL-1105 AZ Amsterdam, Netherlands; Erasmus Med Ctr Rotterdam, Inst Epidemiol & Biostat, Rotterdam, Netherlands
COUNTRY OF AUTHOR: Netherlands

SOURCE: AMERICAN JOURNAL OF MEDICAL GENETICS PART A, (1 MAY 2004) Vol. 126A, No. 4, pp. 403-412. ISSN: 0148-7299.
PUBLISHER: WILEY-LISS, DIV JOHN WILEY & SONS INC, 605 THIRD AVE, NEW YORK, NY 10158-0012 USA.
DOCUMENT TYPE: Article; Journal
LANGUAGE: English
REFERENCE COUNT: 44
ENTRY DATE: Entered STN: 21 May 2004
Last Updated on STN: 21 May 2004
ABSTRACT IS AVAILABLE IN THE ALL AND IALL FORMATS

AB Pseudoxanthoma elasticum (PXE) is a progressive disorder of elastic fibers in skin, eyes, and arterial walls. It is caused by mutations in the ABCC6 gene. Most patients are sporadic cases. The majority of familial cases show autosomal recessive (AR) inheritance, but autosomal dominant (AD) inheritance has also been reported. We reviewed the literature on AD PXE and we studied in detail, both clinically and by DNA studies, a selection of potentially AD pedigrees from our patient population consisting of 59 probands and their family members. Individuals were considered to have definite PXE if they had two of the following three criteria: characteristic ophthalmologic signs, characteristic dermatologic signs, and a positive skin biopsy. In the literature we found only three families with definite PXE in two successive generations and no families with definite PXE in three or more generations. Our own data set comprised three putative AD families. Extensive DNA studies revealed a mutation in only one ABCC6 allele in the patients of these families. Only one of our families showed definite PXE in two generations. Linkage studies revealed that pseudo-dominance was unlikely in this family. In the other two families AD PXE could not be confirmed after extensive clinical examinations and application of our criteria, since definite PXE was not present in two or more generations. Conclusion: the inheritance pattern in PXE usually is AR. Part of the phenotype in family members of PXE patients might be due to expression in heterozygous carriers of an AR disease. AD inheritance in PXE may exist, but is both after careful literature study and in our patient material much rarer than previously thought. (C) 2003 Wiley-Liss, Inc.

L4 ANSWER 11 OF 22 MEDLINE on STN DUPLICATE 7
ACCESSION NUMBER: 2005096903 MEDLINE
DOCUMENT NUMBER: PubMed ID: 15727254
TITLE: Efficient molecular diagnostic strategy for ABCC6 in pseudoxanthoma elasticum.
AUTHOR: Hu Xiaofeng; Plomp Astrid; Gorgels Theo; Brink Jacoline Ten; Loves Willem; Mannens Marcel; de Jong Paulus T V M; Bergen Arthur A B
CORPORATE SOURCE: Netherlands Ophthalmic Research Institute, KNAW, Amsterdam, The Netherlands.
SOURCE: Genetic testing, (2004 Fall) Vol. 8, No. 3, pp. 292-300. Journal code: 9802546. ISSN: 1090-6576.
PUB. COUNTRY: United States
DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE)
LANGUAGE: English
FILE SEGMENT: Priority Journals
ENTRY MONTH: 200504
ENTRY DATE: Entered STN: 1 Mar 2005
Last Updated on STN: 2 Apr 2005
Entered Medline: 1 Apr 2005

AB Pseudoxanthoma elasticum (PXE) is a hereditary disorder of connective tissue with skin, cardiovascular, and visual involvement. In familial cases, PXE usually segregates in an autosomal recessive fashion. The aim of this manuscript is to describe an efficient strategy for DNA diagnosis of PXE. The two most

frequent mutations, R1141X and an ABCC6 del exons 23-29, as well as a core set of mutations, were identified by restriction enzyme digestion and size separation on agarose gels. Next, in the remaining patient group in which only one or no mutant allele was found, the complete coding sequence was analyzed using denaturing high-performance liquid chromatography (dHPLC). All variations found were confirmed by direct DNA sequencing. Finally, Southern blot was used to investigate the potential presence of small or large deletions. Twenty different mutations, including two novel mutations in the ABCC6 gene, were identified in 80.3% of the 76 patients, and 58.6% of the 152 ABCC6 alleles analyzed. With this strategy, 70 (78.7%) out of 89 mutant alleles could be detected within a week. We conclude that this strategy leads to both reliable and time-saving screening for mutations in the ABCC6 gene in sporadic cases and in families with PXE.

L4 ANSWER 12 OF 22 BIOSIS COPYRIGHT (c) 2007 The Thomson Corporation on STN
ACCESSION NUMBER: 2005:319112 BIOSIS
DOCUMENT NUMBER: PREV200510114507
TITLE: Genotype-phenotype correlation in 62 patients with pseudoxanthoma elasticum.
AUTHOR(S): Fuchsel, L. [Reprint Author]; Kozic, H.; McGuigan, K.; Skvarka, C.; Jacobson, M.; Uitto, J.; Ringpfeil, F.
CORPORATE SOURCE: Jefferson Med Coll, Philadelphia, PA USA
SOURCE: Journal of Investigative Dermatology, (MAR 2004) Vol. 122, No. 3, pp. A93.
Meeting Info.: 65th Annual Meeting of the Society-for-Investigative-Dermatology. Providence, RI, USA. April 28 -May 01, 2004. Soc Investigat Dermatol.
CODEN: JIDEAE. ISSN: 0022-202X.
DOCUMENT TYPE: Conference; (Meeting)
Conference; Abstract; (Meeting Abstract)
LANGUAGE: English
ENTRY DATE: Entered STN: 25 Aug 2005
Last Updated on STN: 25 Aug 2005

L4 ANSWER 13 OF 22 BIOSIS COPYRIGHT (c) 2007 The Thomson Corporation on STN
ACCESSION NUMBER: 2005:319073 BIOSIS
DOCUMENT NUMBER: PREV200510114468
TITLE: dHPLC screening detects novel and recurrent mutations in pseudoxanthoma elasticum.
AUTHOR(S): Fratta, S. [Reprint Author]; Ringpfeil, F.; Terry, S.; Terry, P.; Uitto, J.; Pfendner, E. G.
CORPORATE SOURCE: Thomas Jefferson Univ, Philadelphia, PA 19107 USA
SOURCE: Journal of Investigative Dermatology, (MAR 2004) Vol. 122, No. 3, pp. A87,A86.
Meeting Info.: 65th Annual Meeting of the Society-for-Investigative-Dermatology. Providence, RI, USA. April 28 -May 01, 2004. Soc Investigat Dermatol.
CODEN: JIDEAE. ISSN: 0022-202X.
DOCUMENT TYPE: Conference; (Meeting)
Conference; Abstract; (Meeting Abstract)
LANGUAGE: English
ENTRY DATE: Entered STN: 25 Aug 2005
Last Updated on STN: 25 Aug 2005

L4 ANSWER 14 OF 22 MEDLINE on STN DUPLICATE 8
ACCESSION NUMBER: 2003200483 MEDLINE
DOCUMENT NUMBER: PubMed ID: 12714611
TITLE: Analysis of the frequent R1141X mutation in the ABCC6 gene in pseudoxanthoma elasticum.
AUTHOR: Hu Xiaofeng; Peek Ron; Plomp Astrid; ten Brink Jacoline

ten; Scheffer George; van Soest Simone; Leys Anita; de Jong Paulus T V M; Bergen Arthur A B
CORPORATE SOURCE: Netherlands Ophthalmic Research Institute, Royal Netherlands Academy of Art and Sciences (KNAW), Amsterdam, The Netherlands.
SOURCE: Investigative ophthalmology & visual science, (2003 May) Vol. 44, No. 5, pp. 1824-9.
Journal code: 7703701. ISSN: 0146-0404.
PUB. COUNTRY: United States
DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE)
LANGUAGE: English
FILE SEGMENT: Priority Journals
ENTRY MONTH: 200305
ENTRY DATE: Entered STN: 1 May 2003
Last Updated on STN: 20 May 2003
Entered Medline: 19 May 2003

AB PURPOSE: To characterize the ABCC6 R1141X nonsense mutation, which is implicated in more than 25% of a cohort of patients from The Netherlands with pseudoxanthoma elasticum (PXE). METHODS: A combination of single-strand conformational polymorphism (SSCP), PCR, sequencing, and Southern blot analysis was used to identify mutations in the ABCC6 gene in 62 patients. Haplotypes of 16 patients with the R1141X mutation were determined with eight polymorphic markers spanning the ABCC6 locus. The effect of the R1141X mutation on the expression of ABCC6 was studied in leukocytes and cultured dermal fibroblasts from affected skin in patients heterozygous or homozygous for the R1141X mutation. ABCC6 expression was analyzed by RT-PCR and immunocytochemistry with ABCC6-specific monoclonal antibodies. RESULTS: The ABCC6 R1141X mutation was found on 19 alleles in 16 patients with PXE and occurred in heterozygous, homozygous, or compound heterozygous form. All R1141X alleles were associated with a common haplotype, covering at least three intragenic ABCC6 markers. None of the patients or healthy control subjects had a similar ABCC6 haplotype. Furthermore, the results showed that the expression of the normal allele in R1141X heterozygotes was predominant, whereas no detectable, or very low, ABCC6 mRNA levels were found in R1141X homozygotes. Immunocytochemical staining of cultured dermal fibroblasts with ABCC6-specific monoclonal antibodies showed no evidence of the presence of a truncated protein in patients with PXE who were homozygous for R1141X. CONCLUSIONS: A specific founder effect for the R1141X mutation exists in Dutch patients with PXE. The R1141X mutation induces instability of the aberrant mRNA. Functional haploinsufficiency or loss of function of ABCC6 caused by mechanisms, such as nonsense-mediated decay (NMD), may be involved in the PXE phenotype.

L4 ANSWER 15 OF 22 MEDLINE on STN DUPLICATE 9
ACCESSION NUMBER: 2003587243 MEDLINE
DOCUMENT NUMBER: PubMed ID: 14667841
TITLE: Multidrug resistance protein-6 (MRP6) in human dermal fibroblasts. Comparison between cells from normal subjects and from Pseudoxanthoma elasticum patients.
AUTHOR: Boraldi F; Quaglino D; Croce M A; Garcia Fernandez M I; Tiozzo R; Gheduzzi D; Bacchelli B; Pasquali Ronchetti I
CORPORATE SOURCE: Department of Biomedical Sciences, University of Modena and Reggio Emilia, via Campi 287, 41100 Modena, Italy.
SOURCE: Matrix biology : journal of the International Society for Matrix Biology, (2003 Nov) Vol. 22, No. 6, pp. 491-500.
Journal code: 9432592. ISSN: 0945-053X.
PUB. COUNTRY: Germany: Germany, Federal Republic of
DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE)

LANGUAGE: English
FILE SEGMENT: Priority Journals
ENTRY MONTH: 200410
ENTRY DATE: Entered STN: 16 Dec 2003
Last Updated on STN: 7 Oct 2004
Entered Medline: 6 Oct 2004

AB Multidrug resistance protein-6 (MRP6) is a membrane transporter whose deficiency leads to the connective tissue disorder Pseudoxanthoma elasticum (PXE). In vitro dermal fibroblasts from normal and PXE subjects, homozygous for the R1141X mutation, were compared for their ability to accumulate and to release fluorescent calcein, in the absence and in the presence of inhibitors and competitors of the MDR-multidrug resistance protein (MRP) systems, such as 3-(3-(2-(7-choro-2 quinolinyl) ethenyl)phenyl ((3-dimethyl amino-3-oxo-propyl)thio) methyl) propanoic acid (MK571), verapamil (VPL), vinblastine (VBL), chlorambucil (CHB), benzbromarone (BNZ) and indomethacin (IDM). In the absence of chemicals, calcein accumulation was significantly higher and the release significantly slower in PXE cells compared to controls. VBL and CHB reduced calcein release in both cell strains, without affecting the differences between PXE and control fibroblasts. VPL, BNZ and IDM consistently delayed calcein release from both control and PXE cells; moreover, they abolished the differences between normal and MRP6 -deficient fibroblasts observed in the absence of chemicals. These findings suggest that VPL, BNZ and IDM interfere with MRP6 -dependent calcein extrusion in in vitro human normal fibroblasts. Interestingly, MK571 almost completely abolished calcein release from PXE cells, whereas it induced a strong but less complete inhibition in control fibroblasts, suggesting that MRP6 is not inhibited by MK571. Data show that MRP6 is active in human fibroblasts, and that its sensitivity to inhibitors and competitors of MDR-MRPs' membrane transporters is different from that of other translocators, namely, MRP1. It could be suggested that MRP1 and MRP6 transport different physiological substances and that MRP6 deficiency cannot be overcome by other membrane transporters, at least in fibroblasts. These data further support the hypothesis that MRP6 deficiency may be relevant for fibroblast metabolism and responsible for the metabolic alterations of these cells at the basis of connective tissue clinical manifestations of PXE.

L4 ANSWER 16 OF 22 MEDLINE on STN DUPLICATE 10
ACCESSION NUMBER: 2003157019 MEDLINE
DOCUMENT NUMBER: PubMed ID: 12673275
TITLE: ABCC6/MRP6 mutations: further insight into the molecular pathology of pseudoxanthoma elasticum.
AUTHOR: Hu Xiaofeng; Plomp Astrid; Wijnholds Jan; Ten Brink Jacoline; van Soest Simone; van den Born L Ingeborgh; Leys Anita; Peek Ron; de Jong Paulus T V M; Bergen Arthur A B
CORPORATE SOURCE: Netherlands Ophthalmic Research Institute, KNAW, Amsterdam, The Netherlands.
SOURCE: European journal of human genetics : EJHG, (2003 Mar) Vol. 11, No. 3, pp. 215-24.
Journal code: 9302235. ISSN: 1018-4813.
PUB. COUNTRY: England: United Kingdom
DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE)
LANGUAGE: English
FILE SEGMENT: Priority Journals
OTHER SOURCE: GENBANK-Q92878; REFSEQ-NP_000341; REFSEQ-NP_001162; REFSEQ-XP_004980
ENTRY MONTH: 200311
ENTRY DATE: Entered STN: 4 Apr 2003
Last Updated on STN: 5 Nov 2003
Entered Medline: 4 Nov 2003

AB Pseudoxanthoma elasticum (PXE) is a hereditary disease characterized by progressive dystrophic mineralization of the elastic fibres. PXE patients frequently present with skin lesions and visual acuity loss. Recently, we and others showed that PXE is caused by mutations in the ABCC6/MRP6 gene. However, the molecular pathology of PXE is complicated by yet unknown factors causing the variable clinical expression of the disease. In addition, the presence of ABCC6/MRP6 pseudogenes and multiple ABCC6/MRP6-associated deletions complicate interpretation of molecular genetic studies. In this study, we present the mutation spectrum of ABCC6/MRP6 in 59 PXE patients from the Netherlands. We detected 17 different mutations in 65 alleles. The majority of mutations occurred in the NBF1 (nucleotide binding fold) domain, in the eighth cytoplasmatic loop between the 15th and 16th transmembrane regions, and in NBF2 of the predicted ABCC6 /MRP6 protein. The R1141X mutation was by far the most common mutation identified in 19 (32.2%) patients. The second most frequent mutation, an intragenic deletion from exon 23 to exon 29 in ABCC6/MRP6, was detected in 11 (18.6%) of the patients. Our data include 11 novel ABCC6/MRP6 mutations, as well as additional segregation data relevant to the molecular pathology of PXE in a limited number of patients and families. The consequences of our data for the molecular pathology of PXE are discussed.

L4 ANSWER 17 OF 22 MEDLINE on STN DUPLICATE 11

ACCESSION NUMBER: 2002421555 MEDLINE
DOCUMENT NUMBER: PubMed ID: 12176944
TITLE: Frequent mutation in the ABCC6 gene (R1141X) is associated with a strong increase in the prevalence of coronary artery disease.
AUTHOR: Trip Mieke D; Smulders Yvo M; Wegman Jurgen J; Hu Xiaofeng; Boer Jolanda M A; ten Brink Jacoline B; Zwinderman Aeilko H; Kastelein John J P; Feskens Edith J M; Bergen Arthur A B
CORPORATE SOURCE: Department of Cardiology, Academic Medical Centre, University of Amsterdam, Amsterdam, The Netherlands.. M.D.Trip@AMC.UVA.NL
SOURCE: Circulation, (2002 Aug 13) Vol. 106, No. 7, pp. 773-5. Journal code: 0147763. E-ISSN: 1524-4539.
PUB. COUNTRY: United States
DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE)
LANGUAGE: English
FILE SEGMENT: Abridged Index Medicus Journals; Priority Journals
ENTRY MONTH: 200210
ENTRY DATE: Entered STN: 15 Aug 2002
Last Updated on STN: 17 Oct 2002
Entered Medline: 16 Oct 2002

AB BACKGROUND: Pseudoxanthoma elasticum (PXE) is an inborn disorder of the connective tissue with specific skin, ocular, and cardiovascular disease (CVD) manifestations. Recently, we and others have identified mutations in the gene coding for the ABCC6 transporter in PXE patients with ocular and skin involvement. In the Netherlands, as in the rest of Europe, a particular premature truncation variant ABCC6 (R1141X) was found in a large cohort of PXE patients. Given the association between CVD and PXE, we hypothesized that heterozygosity of this ABCC6 mutation could also confer an increased risk for CVD. METHODS AND RESULTS: To assess the relationship between the frequent R1141X mutation in the ABCC6 gene and the prevalence of premature coronary artery disease (CAD), we conducted a case-control study of 441 patients under the age of 50 years who had definite CAD and 1057 age- and sex-matched population-based controls who were free of coronary disease. Strikingly, the prevalence of the R1141X mutation was 4.2 times higher among patients than among controls (3.2% versus 0.8%; P<0.001).

Consequently, among subjects with the R1141X mutation, the odds ratio for a coronary event was 4.23 (95% CI: 1.76 to 10.20, P= 0.001). CONCLUSION: The presence of the R1141X mutation in the ABCC6 gene is associated with a sharply increased risk of premature CAD.

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ACCESSION NUMBER: 2003:165025 BIOSIS
DOCUMENT NUMBER: PREV200300165025
TITLE: Molecular analysis of Pseudoxanthoma Elasticum: spectrum of ABCC6 gene mutations in the Netherlands.
AUTHOR(S): Hu, X. [Reprint Author]; Plomp, A. [Reprint Author]; Ten Brink, J. B. [Reprint Author]; Wijnholds, J. [Reprint Author]; Schuurman, E. J. [Reprint Author]; Soest, S. van [Reprint Author]; Oud, M. [Reprint Author]; Peek, R. [Reprint Author]; Jong, P. T. V. M. [Reprint Author]; Bergen, A. A. B. [Reprint Author]
CORPORATE SOURCE: Research Unit Ophthalmogenetics, Netherlands Ophthalmic Research Institute, Amsterdam, Netherlands
SOURCE: ARVO Annual Meeting Abstract Search and Program Planner, (2002) Vol. 2002, pp. Abstract No. 2394. cd-rom. Meeting Info.: Annual Meeting of the Association For Research in Vision and Ophthalmology. Fort Lauderdale, Florida, USA. May 05-10, 2002.
DOCUMENT TYPE: Conference; (Meeting)
Conference; Abstract; (Meeting Abstract)
LANGUAGE: English
ENTRY DATE: Entered STN: 2 Apr 2003
Last Updated on STN: 2 Apr 2003

AB Purpose: To better understand the function of ABCC6 in the pathogenesis of Pseudoxanthoma Elasticum(PXE) and to direct the service of clinical geneticists for PXE patients by offering the relationship between genotype and phenotype. Methods: The clinical diagnosis of PXE in individuals met all criteria reported by the PXE consensus conference in 1994. The majority of patients are of Dutch descent and were primarily ascertained through the national register of genetic eye diseases at the Netherlands Ophthalmic Research Institute. The ABCC6 gene was screened in 57 unrelated familial and sporadic PXE cases by single strand conformation polymorphism (SSCP), sequencing analysis, and Southern blot. Results: We identified 45 mutation carriers with at least one disease-causing allele, representing 79% of PXE patients studied. A total of 15 different mutations were characterized. All are likely to be causative mutations since by were excluded from 200 control chromosomes. All classes of mutation were detected, including nonsense, missense, frameshift, and splice site mutations. The most mutations create stop codons in the gene, as a consequence, either a shorter mRNA or a truncated protein. Whereas most mutations occur only once, the nonsense mutation R1141X (a C-to-T substitution) within exon 24 accounts for 15 of 45 (33%) of all mutations detected. Seven different missense mutations were found in 10 patients. Four different frameshifting insertions/deletions and one splice site mutation were identified in 15 patients. A deletion spanning exon 23 to 29 in ABCC6 gene was detected in 3 unrelated families. One patient showed compound heterozygous deletions, combining an intragenic exon 23-29 deletion and large intergenic deletion which encompasses ABCC1, ABCC6 and MYH11. Conclusions: Multiple mutations in the ABCC6 gene are associated with PXE. A scan of the entire coding sequencing and duplication part of the gene may be required to detect the causative mutation in PXE patients. It is likely that PXE, in a subset of cases is caused by loss of ABCC6 function.

L4 ANSWER 19 OF 22 MEDLINE on STN DUPLICATE 12
ACCESSION NUMBER: 2001492688 MEDLINE
DOCUMENT NUMBER: PubMed ID: 11536079
TITLE: A spectrum of ABCC6 mutations is responsible for pseudoxanthoma elasticum.
AUTHOR: Le Saux O; Beck K; Sachsinger C; Silvestri C; Treiber C; Goring H H; Johnson E W; De Paepe A; Pope F M; Pasquali-Ronchetti I; Bercovitch L; Marais A S; Viljoen D L; Terry S F; Boyd C D
CORPORATE SOURCE: Pacific Biomedical Research Center, University of Hawai'i, Honolulu, HI 96822, USA.
CONTRACT NUMBER: EY13019 (NEI)
RR03061 (NCRR)
SOURCE: American journal of human genetics, (2001 Oct) Vol. 69, No. 4, pp. 749-64. Electronic Publication: 2001-08-31. Journal code: 0370475. ISSN: 0002-9297.
PUB. COUNTRY: United States
DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE)
LANGUAGE: English
FILE SEGMENT: Priority Journals
OTHER SOURCE: GENBANK-AC002039; GENBANK-AC002045; GENBANK-AC002492; GENBANK-U91318; OMIM-177850; OMIM-264800; REFSEQ-NM_000352; REFSEQ-NM_000492; REFSEQ-NM_000927; REFSEQ-NM_001171; REFSEQ-NT_010393; REFSEQ-XM_017599; REFSEQ-XM_017612
ENTRY MONTH: 200110
ENTRY DATE: Entered STN: 6 Sep 2001
Last Updated on STN: 5 Jan 2003
Entered Medline: 18 Oct 2001

AB To better understand the pathogenetics of pseudoxanthoma elasticum (PXE), we performed a mutational analysis of ATP-binding cassette subfamily C member 6 (ABCC6) in 122 unrelated patients with PXE, the largest cohort of patients yet studied. Thirty-six mutations were characterized, and, among these, 28 were novel variants (for a total of 43 PXE mutations known to date). Twenty-one alleles were missense variants, six were small insertions or deletions, five were nonsense, two were alleles likely to result in aberrant mRNA splicing, and two were large deletions involving ABCC6. Although most mutations appeared to be unique variants, two disease-causing alleles occurred frequently in apparently unrelated individuals. R1141X was found in our patient cohort at a frequency of 18.8% and was preponderant in European patients. ABCC6del23-29 occurred at a frequency of 12.9% and was prevalent in patients from the United States. These results suggested that R1141X and ABCC6del23-29 might have been derived regionally from founder alleles. Putative disease-causing mutations were identified in approximately 64% of the 244 chromosomes studied, and 85.2% of the 122 patients were found to have at least one disease-causing allele. Our results suggest that a fraction of the undetected mutant alleles could be either genomic rearrangements or mutations occurring in noncoding regions of the ABCC6 gene. The distribution pattern of ABCC6 mutations revealed a cluster of disease-causing variants within exons encoding a large C-terminal cytoplasmic loop and in the C-terminal nucleotide-binding domain (NBD2). We discuss the potential structural and functional significance of this mutation pattern within the context of the complex relationship between the PXE phenotype and the function of ABCC6.

L4 ANSWER 20 OF 22 MEDLINE on STN DUPLICATE 13
ACCESSION NUMBER: 2000408767 MEDLINE
DOCUMENT NUMBER: PubMed ID: 10913334
TITLE: Homozygosity for the R1268Q mutation in MRP6, the pseudoxanthoma elasticum gene, is not disease-causing.
AUTHOR: Germain D P; Perdu J; Remones V; Jeunemaitre X

CORPORATE SOURCE: Departement de Genetique, Universite Paris VI, Paris, France.. dominique.germain@brs.ap-hop-paris.fr
SOURCE: Biochemical and biophysical research communications, (2000 Aug 2) Vol. 274, No. 2, pp. 297-301.
Journal code: 0372516. ISSN: 0006-291X.
PUB. COUNTRY: United States
DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE)
LANGUAGE: English
FILE SEGMENT: Priority Journals
ENTRY MONTH: 200008
ENTRY DATE: Entered STN: 1 Sep 2000
Last Updated on STN: 19 Oct 2000
Entered Medline: 24 Aug 2000

AB Pseudoxanthoma elasticum (PXE) is an inherited systemic disorder of connective tissue, characterized by progressive calcification of the elastic fibers in the eye, the skin, and the cardiovascular system, resulting in decreased vision, skin lesions, and life-threatening vascular disease, with highly variable phenotypic expression. The PXE locus has been mapped to chromosome 16p13.1, and was recently further refined to a 500 kb-region, containing two pseudogenes and four candidate genes. In a comprehensive mutational screening, we were able to exclude the responsibility of pM5, UNK, and MRP1 genes, candidate on the basis of their genetic localization. Conversely, we have found pathogenetic mutations in the MRP6 gene, in patients affected with PXE, indicating that human MRP6, which encodes a 1503 amino-acids membrane protein, member of the human ATP binding cassette (ABC) transporters superfamily, is the gene responsible for PXE. In one large PXE pedigree for which we had identified a nonsense mutation (R1141X), we came across a G to A transition at position 3803 of the MRP6 cDNA sequence (R1268Q). Astonishingly, this latter variant was found at the homozygous state in the proband's unaffected husband. We investigated the R1268Q mutation, and found the Q1268 allele at a relatively high frequency (0.19) in a Caucasian control population (n = 62 subjects). Genotype frequencies were in Hardy-Weinberg equilibrium, and three healthy volunteers were homozygous for the Q1268 allele. These data indicate that the R1268Q variant in the MRP6 gene does not cause PXE per se. Further studies will elucidate if it may play a role when found in compound heterozygotes.

L4 ANSWER 21 OF 22 CAPLUS COPYRIGHT 2007 ACS on STN
ACCESSION NUMBER: 2000:397370 CAPLUS
DOCUMENT NUMBER: 133:118224
TITLE: Mutations in ABCC6 cause pseudoxanthoma elasticum
AUTHOR(S): Bergen, Arthur A. B.; Plomp, Astrid S.; Schuurman, Ellen J.; Terry, Sharon; Breuning, Martijn; Dauwerse, Hans; Swart, Jaap; Kool, Marcel; Van Soest, Simone; Baas, Frank; ten Brink, Jacoline B.; De Jong, Paulus T. V. M.
CORPORATE SOURCE: The Netherlands Ophthalmic Research Institute, Amsterdam, Neth.
SOURCE: Nature Genetics (2000), 25(2), 228-231
CODEN: NGENEC; ISSN: 1061-4036
PUBLISHER: Nature America Inc.
DOCUMENT TYPE: Journal
LANGUAGE: English

AB Pseudoxanthoma elasticum (PXE) is a heritable disorder of the connective tissue. PXE patients frequently experience visual field loss and skin lesions, and occasionally cardiovascular complications. Histopathol. findings reveal calcification of the elastic fibers and abnormalities of the collagen fibrils. Most PXE patients are sporadic, but autosomal recessive and dominant inheritance

are also observed The authors previously localized the PXE gene to chromosome 16p13.1 and constructed a phys. map. Here the authors describe homozygosity mapping in five PXE families and the detection of deletions or mutations in ABCC6 (formerly MRP6) associated with all genetic forms of PXE in seven patients or families.
REFERENCE COUNT: 23 THERE ARE 23 CITED REFERENCES AVAILABLE FOR THIS RECORD. ALL CITATIONS AVAILABLE IN THE RE FORMAT

L4 ANSWER 22 OF 22 CAPLUS COPYRIGHT 2007 ACS on STN
ACCESSION NUMBER: 2000:397369 CAPLUS
DOCUMENT NUMBER: 133:118223
TITLE: Mutations in a gene encoding an ABC transporter cause pseudoxanthoma elasticum
AUTHOR(S): Le Saux, Olivier; Urban, Zsolt; Tschuch, Cordula; Csiszar, Katalin; Bacchelli, Barbara; Quaglino, Daniela; Pasquali-Ronchetti, Ivonne; Pope, F. Michael; Richards, Allan; Terry, Sharon; Bercovitch, Lionel; De Paepe, Anne; Boyd, Charles D.
CORPORATE SOURCE: Laboratory of Matrix Pathobiology, Pacific Biomedical Research Center, University of Hawai'i, Honolulu, HI, USA
SOURCE: Nature Genetics (2000), 25(2), 223-227
CODEN: NGENEC; ISSN: 1061-4036
PUBLISHER: Nature America Inc.
DOCUMENT TYPE: Journal
LANGUAGE: English
AB Pseudoxanthoma elasticum (PXE) is a heritable disorder characterized by calcification of elastic fibers in skin, arteries and retina that results in dermal lesions with associated laxity and loss of elasticity, arterial insufficiency and retinal hemorrhages leading to macular degeneration. PXE is usually found as a sporadic disorder, but examples of both autosomal recessive and autosomal dominant forms of PXE have been observed Partial manifestations of the PXE phenotype have also been described in presumed carriers in PXE families. Linkage of both dominant and recessive forms of PXE to a 5-cM domain on chromosome 16p13.1 has been reported. The authors have refined this locus to an 820-kb region containing 6 candidate genes. Here the authors report the exclusion of five of these genes and the identification of the first mutations responsible for the development of PXE in a gene encoding a protein associated with multidrug resistance (ABCC6).
REFERENCE COUNT: 26 THERE ARE 26 CITED REFERENCES AVAILABLE FOR THIS RECORD. ALL CITATIONS AVAILABLE IN THE RE FORMAT

=> d ib l3 1-68
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L3 ANSWER 1 OF 68 MEDLINE on STN
ACCESSION NUMBER: 2006495014 MEDLINE
DOCUMENT NUMBER: PubMed ID: 16835894
TITLE: Mutational analysis of the ABCC6 gene and the proximal ABCC6 gene promoter in German patients with pseudoxanthoma elasticum (PXE).
AUTHOR: Schulz Veronika; Hendig Doris; Henjakovic Maja; Szliska Christiane; Kleesiek Knut; Gotting Christian
CORPORATE SOURCE: Institut fur Laboratoriums- und Transfusionsmedizin, Herz- und Diabeteszentrum Nordrhein-Westfalen, Universitatsklinik der Ruhr-Universitat Bochum, Bad Oeynhausen, Germany.
SOURCE: Human mutation, (2006 Aug) Vol. 27, No. 8, pp. 831.
Journal code: 9215429. E-ISSN: 1098-1004.
PUB. COUNTRY: United States
DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE)
LANGUAGE: English
FILE SEGMENT: Priority Journals
ENTRY MONTH: 200610
ENTRY DATE: Entered STN: 22 Aug 2006
Last Updated on STN: 17 Oct 2006
Entered Medline: 16 Oct 2006

L3 ANSWER 2 OF 68 MEDLINE on STN
ACCESSION NUMBER: 2006064186 MEDLINE
DOCUMENT NUMBER: PubMed ID: 16384891
TITLE: Role of serum fetuin-A, a major inhibitor of systemic calcification, in pseudoxanthoma elasticum.
AUTHOR: Hendig Doris; Schulz Veronika; Arndt Marius; Szliska Christiane; Kleesiek Knut; Gotting Christian
CORPORATE SOURCE: Institut fur Laboratoriums- und Transfusionsmedizin, Herz- und Diabeteszentrum Nordrhein-Westfalen, Universitatsklinik der Ruhr-Universitat Bochum, Bad Oeynhausen, Germany.
SOURCE: Clinical chemistry, (2006 Feb) Vol. 52, No. 2, pp. 227-34.
Electronic Publication: 2005-12-29.
Journal code: 9421549. ISSN: 0009-9147.
PUB. COUNTRY: United States
DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE)
LANGUAGE: English
FILE SEGMENT: Priority Journals
ENTRY MONTH: 200603
ENTRY DATE: Entered STN: 2 Feb 2006
Last Updated on STN: 11 Mar 2006
Entered Medline: 10 Mar 2006

L3 ANSWER 3 OF 68 MEDLINE on STN
ACCESSION NUMBER: 2006006847 MEDLINE
DOCUMENT NUMBER: PubMed ID: 16392638
TITLE: Novel mutations in the ABCC6 gene of German patients with pseudoxanthoma elasticum.
AUTHOR: Schulz Veronika; Hendig Doris; Szliska Christiane; Gotting Christian; Kleesiek Knut
CORPORATE SOURCE: Institut fur Laboratoriums- und Transfusionsmedizin, Herz- und Diabeteszentrum Nordrhein-Westfalen, Universitatsklinik der Ruhr-Universitat Bochum, Georgstrasse 11, 32545 Bad Oeynhausen, Germany.
SOURCE: Human biology; an international record of research, (2005 Jun) Vol. 77, No. 3, pp. 367-84.
Journal code: 0116717. ISSN: 0018-7143.
PUB. COUNTRY: United States
DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE)
LANGUAGE: English
FILE SEGMENT: Priority Journals
ENTRY MONTH: 200602
ENTRY DATE: Entered STN: 6 Jan 2006

Last Updated on STN: 22 Feb 2006
Entered Medline: 21 Feb 2006

L3 ANSWER 4 OF 68 MEDLINE on STN
ACCESSION NUMBER: 2005641044 MEDLINE
DOCUMENT NUMBER: PubMed ID: 16133423
TITLE: Elevated xylosyltransferase I activities in pseudoxanthoma elasticum (PXE) patients as a marker of stimulated proteoglycan biosynthesis.
AUTHOR: Gotting Christian; Hendig Doris; Adam Alexandra; Schon Sylvia; Schulz Veronika; Szliska Christiane; Kuhn Joachim; Kleesiek Knut
CORPORATE SOURCE: Institut fur Laboratoriums-und Transfusionsmedizin, Herz-und Diabeteszentrum Nordrhein-Westfalen, Universitatsklinik der Ruhr-Universitat Bochum, Georgstrasse 11, 32545 Bad Oeynhausen, Germany.. cgoetting@hdz-nrw.de
SOURCE: Journal of molecular medicine (Berlin, Germany), (2005 Dec) Vol. 83, No. 12, pp. 984-92. Electronic Publication: 2005-08-24.
Journal code: 9504370. ISSN: 0946-2716.
PUB. COUNTRY: Germany: Germany, Federal Republic of
DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE)
LANGUAGE: English
FILE SEGMENT: Priority Journals
ENTRY MONTH: 200605
ENTRY DATE: Entered STN: 3 Dec 2005
Last Updated on STN: 4 May 2006
Entered Medline: 3 May 2006

L3 ANSWER 5 OF 68 MEDLINE on STN
ACCESSION NUMBER: 2005202853 MEDLINE
DOCUMENT NUMBER: PubMed ID: 15837081
TITLE: Patients with premature coronary artery disease who carry the ABCC6 R1141X mutation have no Pseudoxanthoma Elasticum phenotype.
AUTHOR: Wegman Jurgen J; Hu Xiaofeng; Tan Hendra; Bergen Arthur A B; Trip Mieke D; Kastelein John J P; Smulders Yvo M
CORPORATE SOURCE: Department of Vascular Medicine, Academic Medical Center, University of Amsterdam, The Netherlands.
SOURCE: International journal of cardiology, (2005 Apr 28) Vol. 100, No. 3, pp. 389-93.
Journal code: 8200291. ISSN: 0167-5273.
PUB. COUNTRY: Ireland
DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE)
LANGUAGE: English
FILE SEGMENT: Priority Journals
ENTRY MONTH: 200507
ENTRY DATE: Entered STN: 20 Apr 2005
Last Updated on STN: 29 Jul 2005
Entered Medline: 28 Jul 2005

L3 ANSWER 6 OF 68 MEDLINE on STN
ACCESSION NUMBER: 2005096903 MEDLINE
DOCUMENT NUMBER: PubMed ID: 15727254
TITLE: Efficient molecular diagnostic strategy for ABCC6 in pseudoxanthoma elasticum.
AUTHOR: Hu Xiaofeng; Plomp Astrid; Gorgels Theo; Brink Jacoline Ten; Loves Willem; Mannens Marcel; de Jong Paulus T V M; Bergen Arthur A B
CORPORATE SOURCE: Netherlands Ophthalmic Research Institute, KNAW, Amsterdam, The Netherlands.
SOURCE: Genetic testing, (2004 Fall) Vol. 8, No. 3, pp. 292-300.
Journal code: 9802546. ISSN: 1090-6576.

PUB. COUNTRY: United States
DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE)
LANGUAGE: English
FILE SEGMENT: Priority Journals
ENTRY MONTH: 200504
ENTRY DATE: Entered STN: 1 Mar 2005
Last Updated on STN: 2 Apr 2005
Entered Medline: 1 Apr 2005

L3 ANSWER 7 OF 68 MEDLINE on STN
ACCESSION NUMBER: 2004471837 MEDLINE
DOCUMENT NUMBER: PubMed ID: 15382558
TITLE: [From gene to disease; pseudoxanthoma elasticum and the ABCC6 gene].
Van gen naar ziekte; pseudoxanthoma elasticum en het ABCC6-gen.
AUTHOR: Bergen A A B; Plomp A S; Gorgels T G M F; de Jong P T V M
CORPORATE SOURCE: Interuniversitair Oogheelkundig Instituut, Meibergdreef 47, 11o05 BA Amsterdam.. a.bergen@ioi.knaw.nl
SOURCE: Nederlands tijdschrift voor geneeskunde, (2004 Aug 7) Vol. 148, No. 32, pp. 1586-9. Ref: 3
Journal code: 0400770. ISSN: 0028-2162.
PUB. COUNTRY: Netherlands
DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE)
General Review; (REVIEW)
LANGUAGE: Dutch
FILE SEGMENT: Priority Journals
ENTRY MONTH: 200410
ENTRY DATE: Entered STN: 23 Sep 2004
Last Updated on STN: 13 Oct 2004
Entered Medline: 12 Oct 2004

L3 ANSWER 8 OF 68 MEDLINE on STN
ACCESSION NUMBER: 2004191545 MEDLINE
DOCUMENT NUMBER: PubMed ID: 15086542
TITLE: Novel ABCC6 mutations in pseudoxanthoma elasticum.
AUTHOR: Chassaing Nicolas; Martin Ludovic; Mazereeuw Juliette; Barrie Laurence; Nizard Sonia; Bonafe Jean-Louis; Calvas Patrick; Hovnanian Alain
CORPORATE SOURCE: Department of Medical Genetics, Purpan Hospital, Toulouse, France.
SOURCE: The Journal of investigative dermatology, (2004 Mar) Vol. 122, No. 3, pp. 608-13.
Journal code: 0426720. ISSN: 0022-202X.
PUB. COUNTRY: United States
DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE)
LANGUAGE: English
FILE SEGMENT: Priority Journals
ENTRY MONTH: 200405
ENTRY DATE: Entered STN: 17 Apr 2004
Last Updated on STN: 26 May 2004
Entered Medline: 25 May 2004

L3 ANSWER 9 OF 68 MEDLINE on STN
ACCESSION NUMBER: 2003587243 MEDLINE
DOCUMENT NUMBER: PubMed ID: 14667841
TITLE: Multidrug resistance protein-6 (MRP6) in human dermal fibroblasts. Comparison between cells from normal subjects and from Pseudoxanthoma elasticum patients.
AUTHOR: Boraldi F; Quaglino D; Croce M A; Garcia Fernandez M I; Tiozzo R; Gheduzzi D; Bacchelli B; Pasquali Ronchetti I
CORPORATE SOURCE: Department of Biomedical Sciences, University of Modena and

Reggio Emilia, via Campi 287, 41100 Modena, Italy.
SOURCE: Matrix biology : journal of the International Society for Matrix Biology, (2003 Nov) Vol. 22, No. 6, pp. 491-500. Journal code: 9432592. ISSN: 0945-053X.
PUB. COUNTRY: Germany: Germany, Federal Republic of
DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE)
LANGUAGE: English
FILE SEGMENT: Priority Journals
ENTRY MONTH: 200410
ENTRY DATE: Entered STN: 16 Dec 2003
Last Updated on STN: 7 Oct 2004
Entered Medline: 6 Oct 2004

L3 ANSWER 10 OF 68 MEDLINE on STN
ACCESSION NUMBER: 2003200483 MEDLINE
DOCUMENT NUMBER: PubMed ID: 12714611
TITLE: Analysis of the frequent R1141X mutation in the ABCC6 gene in pseudoxanthoma elasticum.
AUTHOR: Hu Xiaofeng; Peek Ron; Plomp Astrid; ten Brink Jacoline ten; Scheffer George; van Soest Simone; Leys Anita; de Jong Paulus T V M; Bergen Arthur A B
CORPORATE SOURCE: Netherlands Ophthalmic Research Institute, Royal Netherlands Academy of Art and Sciences (KNAW), Amsterdam, The Netherlands.
SOURCE: Investigative ophthalmology & visual science, (2003 May) Vol. 44, No. 5, pp. 1824-9. Journal code: 7703701. ISSN: 0146-0404.
PUB. COUNTRY: United States
DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE)
LANGUAGE: English
FILE SEGMENT: Priority Journals
ENTRY MONTH: 200305
ENTRY DATE: Entered STN: 1 May 2003
Last Updated on STN: 20 May 2003
Entered Medline: 19 May 2003

L3 ANSWER 11 OF 68 MEDLINE on STN
ACCESSION NUMBER: 2003157019 MEDLINE
DOCUMENT NUMBER: PubMed ID: 12673275
TITLE: ABCC6/MRP6 mutations: further insight into the molecular pathology of pseudoxanthoma elasticum.
AUTHOR: Hu Xiaofeng; Plomp Astrid; Wijnholds Jan; Ten Brink Jacoline; van Soest Simone; van den Born L Ingeborgh; Leys Anita; Peek Ron; de Jong Paulus T V M; Bergen Arthur A B
CORPORATE SOURCE: Netherlands Ophthalmic Research Institute, KNAW, Amsterdam, The Netherlands.
SOURCE: European journal of human genetics : EJHG, (2003 Mar) Vol. 11, No. 3, pp. 215-24. Journal code: 9302235. ISSN: 1018-4813.
PUB. COUNTRY: England: United Kingdom
DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE)
LANGUAGE: English
FILE SEGMENT: Priority Journals
OTHER SOURCE: GENBANK-Q92878; REFSEQ-NP_000341; REFSEQ-NP_001162; REFSEQ-XP_004980
ENTRY MONTH: 200311
ENTRY DATE: Entered STN: 4 Apr 2003
Last Updated on STN: 5 Nov 2003
Entered Medline: 4 Nov 2003

L3 ANSWER 12 OF 68 MEDLINE on STN
ACCESSION NUMBER: 2002421555 MEDLINE
DOCUMENT NUMBER: PubMed ID: 12176944

TITLE: Frequent mutation in the ABCC6 gene (R1141X) is associated with a strong increase in the prevalence of coronary artery disease.
AUTHOR: Trip Mieke D; Smulders Yvo M; Wegman Jurgen J; Hu Xiaofeng; Boer Jolanda M A; ten Brink Jacoline B; Zwinderman Aeilko H; Kastelein John J P; Feskens Edith J M; Bergen Arthur A B
CORPORATE SOURCE: Department of Cardiology, Academic Medical Centre, University of Amsterdam, Amsterdam, The Netherlands.. M.D.Trip@AMC.UVA.NL
SOURCE: Circulation, (2002 Aug 13) Vol. 106, No. 7, pp. 773-5. Journal code: 0147763. E-ISSN: 1524-4539.
PUB. COUNTRY: United States
DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE)
LANGUAGE: English
FILE SEGMENT: Abridged Index Medicus Journals; Priority Journals
ENTRY MONTH: 200210
ENTRY DATE: Entered STN: 15 Aug 2002
Last Updated on STN: 17 Oct 2002
Entered Medline: 16 Oct 2002

L3 ANSWER 13 OF 68 MEDLINE on STN
ACCESSION NUMBER: 2001492688 MEDLINE
DOCUMENT NUMBER: PubMed ID: 11536079
TITLE: A spectrum of ABCC6 mutations is responsible for pseudoxanthoma elasticum.
AUTHOR: Le Saux O; Beck K; Sachsinger C; Silvestri C; Treiber C; Goring H H; Johnson E W; De Paepe A; Pope F M; Pasquali-Ronchetti I; Bercovitch L; Marais A S; Viljoen D L; Terry S F; Boyd C D
CORPORATE SOURCE: Pacific Biomedical Research Center, University of Hawai'i, Honolulu, HI 96822, USA.
CONTRACT NUMBER: EY13019 (NEI)
RR03061 (NCRR)
SOURCE: American journal of human genetics, (2001 Oct) Vol. 69, No. 4, pp. 749-64. Electronic Publication: 2001-08-31. Journal code: 0370475. ISSN: 0002-9297.
PUB. COUNTRY: United States
DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE)
LANGUAGE: English
FILE SEGMENT: Priority Journals
OTHER SOURCE: GENBANK-AC002039; GENBANK-AC002045; GENBANK-AC002492; GENBANK-U91318; OMIM-177850; OMIM-264800; REFSEQ-NM_000352; REFSEQ-NM_000492; REFSEQ-NM_000927; REFSEQ-NM_001171; REFSEQ-NT_010393; REFSEQ-XM_017599; REFSEQ-XM_017612
ENTRY MONTH: 200110
ENTRY DATE: Entered STN: 6 Sep 2001
Last Updated on STN: 5 Jan 2003
Entered Medline: 18 Oct 2001

L3 ANSWER 14 OF 68 MEDLINE on STN
ACCESSION NUMBER: 2000408767 MEDLINE
DOCUMENT NUMBER: PubMed ID: 10913334
TITLE: Homozygosity for the R1268Q mutation in MRP6, the pseudoxanthoma elasticum gene, is not disease-causing.
AUTHOR: Germain D P; Perdu J; Remones V; Jeunemaitre X
CORPORATE SOURCE: Departement de Genetique, Universite Paris VI, Paris, France.. dominique.germain@brs.ap-hop-paris.fr
SOURCE: Biochemical and biophysical research communications, (2000 Aug 2) Vol. 274, No. 2, pp. 297-301. Journal code: 0372516. ISSN: 0006-291X.
PUB. COUNTRY: United States
DOCUMENT TYPE: Journal; Article; (JOURNAL ARTICLE)
LANGUAGE: English

FILE SEGMENT: Priority Journals
ENTRY MONTH: 200008
ENTRY DATE: Entered STN: 1 Sep 2000
Last Updated on STN: 19 Oct 2000
Entered Medline: 24 Aug 2000

L3 ANSWER 15 OF 68 CAPLUS COPYRIGHT 2007 ACS on STN
ACCESSION NUMBER: 2006:126787 CAPLUS
DOCUMENT NUMBER: 144:309470
TITLE: Role of serum fetuin-A, a major inhibitor of systemic calcification, in pseudoxanthoma elasticum
AUTHOR(S): Hendig, Doris; Schulz, Veronika; Arndt, Marius; Szliska, Christiane; Kleesiek, Knut; Goetting, Christian
CORPORATE SOURCE: Institut fuer Laboratoriums- und Transfusionsmedizin, Herz- und Diabeteszentrum Nordrhein-Westfalen, Universitaetsklinik der Ruhr-Universitaet Bochum, Bad Oeynhausen, Germany
SOURCE: Clinical Chemistry (Washington, DC, United States) (2006), 52(2), 227-234
CODEN: CLCHAU; ISSN: 0009-9147
PUBLISHER: American Association for Clinical Chemistry
DOCUMENT TYPE: Journal
LANGUAGE: English
REFERENCE COUNT: 53 THERE ARE 53 CITED REFERENCES AVAILABLE FOR THIS RECORD. ALL CITATIONS AVAILABLE IN THE RE FORMAT

L3 ANSWER 16 OF 68 CAPLUS COPYRIGHT 2007 ACS on STN
ACCESSION NUMBER: 2005:1277095 CAPLUS
DOCUMENT NUMBER: 144:347891
TITLE: Elevated xylosyltransferase I activities in pseudoxanthoma elasticum (PXE) patients as a marker of stimulated proteoglycan biosynthesis
AUTHOR(S): Goetting, Christian; Hendig, Doris; Adam, Alexandra; Schoen, Sylvia; Schulz, Veronika; Szliska, Christiane; Kuhn, Joachim; Kleesiek, Knut
CORPORATE SOURCE: Institut fuer Laboratoriums-und Transfusionsmedizin, Universitaetsklinik der Ruhr-Universitaet Bochum, Bad Oeynhausen, 32545, Germany
SOURCE: Journal of Molecular Medicine (Heidelberg, Germany) (2005), 83(12), 984-992
CODEN: JMLME8; ISSN: 0946-2716
PUBLISHER: Springer
DOCUMENT TYPE: Journal
LANGUAGE: English
REFERENCE COUNT: 53 THERE ARE 53 CITED REFERENCES AVAILABLE FOR THIS RECORD. ALL CITATIONS AVAILABLE IN THE RE FORMAT

L3 ANSWER 17 OF 68 CAPLUS COPYRIGHT 2007 ACS on STN
ACCESSION NUMBER: 2005:1107490 CAPLUS
DOCUMENT NUMBER: 144:126802
TITLE: Molecular genetics of pseudoxanthoma elasticum: type and frequency of mutations in ABCC6
AUTHOR(S): Miksch, Sara; Lumsden, Amanda; Guenther, Ulf P.; Foernzler, Dorothee; Christen-Zaech, Stephanie; Daugherty, Carol; Ramesar, Rajkumar S.; Lebwohl, Mark; Hohl, Daniel; Neldner, Kenneth H.; Lindpaintner, Klaus; Richards, Robert I.; Struk, Berthold
CORPORATE SOURCE: Charite, Franz Volhard Clinic, HELIOS Klinikum, Humboldt University Berlin, Germany
SOURCE: Human Mutation (2005), 26(3), 235-248
CODEN: HUMUE3; ISSN: 1059-7794

PUBLISHER: Wiley-Liss, Inc.
DOCUMENT TYPE: Journal
LANGUAGE: English
REFERENCE COUNT: 46 THERE ARE 46 CITED REFERENCES AVAILABLE FOR THIS RECORD. ALL CITATIONS AVAILABLE IN THE RE FORMAT

L3 ANSWER 18 OF 68 CAPLUS COPYRIGHT 2007 ACS on STN
ACCESSION NUMBER: 2005:90130 CAPLUS
DOCUMENT NUMBER: 143:57937
TITLE: ABCC6 mutations in Italian families affected by pseudoxanthoma elasticum (PXE)
AUTHOR(S): Gheduzzi, Dealba; Giudetti, Rita; Anzivino, Claudia; Tarugi, Patrizia; di Leo, Enza; Quaglino, Daniela; Ronchetti, Ivonne Pasquali
CORPORATE SOURCE: Dept. Biomedical Sciences, University of Modena and Reggio Emilia, Modena, Italy
SOURCE: Human Mutation (2004), 24(5), 755/1-755/10
CODEN: HUMUE3; ISSN: 1059-7794
PUBLISHER: Wiley-Liss, Inc.
DOCUMENT TYPE: Journal
LANGUAGE: English
REFERENCE COUNT: 31 THERE ARE 31 CITED REFERENCES AVAILABLE FOR THIS RECORD. ALL CITATIONS AVAILABLE IN THE RE FORMAT

L3 ANSWER 19 OF 68 CAPLUS COPYRIGHT 2007 ACS on STN
ACCESSION NUMBER: 2004:1055593 CAPLUS
DOCUMENT NUMBER: 142:442431
TITLE: Efficient Molecular Diagnostic Strategy for ABCC6 in Pseudoxanthoma Elasticum
AUTHOR(S): Hu, Xiaofeng; Plomp, Astrid; Gorgels, Theo; Ten Brink, Jacoline; Loves, Willem; Mannens, Marcel; de Jong, Paulus T. V. M.; Bergen, Arthur A. B.
CORPORATE SOURCE: Netherlands Ophthalmic Research Institute, KNAW, Amsterdam, Neth.
SOURCE: Genetic Testing (2004), 8(3), 292-300
CODEN: GETEF4; ISSN: 1090-6576
PUBLISHER: Mary Ann Liebert, Inc.
DOCUMENT TYPE: Journal
LANGUAGE: English
REFERENCE COUNT: 24 THERE ARE 24 CITED REFERENCES AVAILABLE FOR THIS RECORD. ALL CITATIONS AVAILABLE IN THE RE FORMAT

L3 ANSWER 20 OF 68 CAPLUS COPYRIGHT 2007 ACS on STN
ACCESSION NUMBER: 2004:320494 CAPLUS
DOCUMENT NUMBER: 140:421614
TITLE: Novel ABCC6 mutations in pseudoxanthoma elasticum
AUTHOR(S): Chassaing, Nicolas; Martin, Ludovic; Mazereeuw, Juliette; Barrie, Laurence; Nizard, Sonia; Bonafe, Jean-Louis; Calvas, Patrick; Hovnanian, Alain
CORPORATE SOURCE: Department of Medical Genetics, INSERM U563, Purpan Hospital, Toulouse, Fr.
SOURCE: Journal of Investigative Dermatology (2004), 122(3), 608-613
CODEN: JIDEAE; ISSN: 0022-202X
PUBLISHER: Blackwell Publishing, Inc.
DOCUMENT TYPE: Journal
LANGUAGE: English
REFERENCE COUNT: 26 THERE ARE 26 CITED REFERENCES AVAILABLE FOR THIS RECORD. ALL CITATIONS AVAILABLE IN THE RE FORMAT

L3 ANSWER 21 OF 68 CAPLUS COPYRIGHT 2007 ACS on STN
ACCESSION NUMBER: 2003:965963 CAPLUS
DOCUMENT NUMBER: 140:143591

TITLE: Multidrug resistance protein-6 (MRP6) in human dermal fibroblasts. Comparison between cells from normal subjects and from Pseudoxanthoma elasticum patients
AUTHOR(S): Boraldi, F.; Quaglino, D.; Croce, M. A.; Garcia Fernandez, M. I.; Tiozzo, R.; Gheduzzi, D.; Bacchelli, B.; Pasquali Ronchetti, I.
CORPORATE SOURCE: Department of Biomedical Sciences, University of Modena and Reggio Emilia, Modena, 41100, Italy
SOURCE: Matrix Biology (2003), 22(6), 491-500
CODEN: MTBOEC; ISSN: 0945-053X
PUBLISHER: Elsevier Science B.V.
DOCUMENT TYPE: Journal
LANGUAGE: English
REFERENCE COUNT: 35 THERE ARE 35 CITED REFERENCES AVAILABLE FOR THIS RECORD. ALL CITATIONS AVAILABLE IN THE RE FORMAT

L3 ANSWER 22 OF 68 CAPLUS COPYRIGHT 2007 ACS on STN
ACCESSION NUMBER: 2003:256353 CAPLUS
DOCUMENT NUMBER: 139:66960
TITLE: ABCC6/MRP6 mutations: further insight into the molecular pathology of pseudoxanthoma elasticum
AUTHOR(S): Hu, Xiaofeng; Plomp, Astrid; Wijnholds, Jan; ten Brink, Jacoline; van Soest, Simone; van den Born, L. Ingeborgh; Leys, Anita; Peek, Ron; de Jong, Paulus T. V. M.; Bergen, Arthur A. B.
CORPORATE SOURCE: Netherlands Ophthalmic Research Institute, Amsterdam, Neth.
SOURCE: European Journal of Human Genetics (2003), 11(3), 215-224
CODEN: EJHGEU; ISSN: 1018-4813
PUBLISHER: Nature Publishing Group
DOCUMENT TYPE: Journal
LANGUAGE: English
REFERENCE COUNT: 41 THERE ARE 41 CITED REFERENCES AVAILABLE FOR THIS RECORD. ALL CITATIONS AVAILABLE IN THE RE FORMAT

L3 ANSWER 23 OF 68 CAPLUS COPYRIGHT 2007 ACS on STN
ACCESSION NUMBER: 2002:703867 CAPLUS
DOCUMENT NUMBER: 138:3050
TITLE: Frequent mutation in the ABCC6 gene (R1141X) is associated with a strong increase in the prevalence of coronary artery disease
AUTHOR(S): Trip, Mieke D.; Smulders, Yvo M.; Wegman, Jurgen J.; Hu, Xiaofeng; Boer, Jolanda M. A.; ten Brink, Jacoline B.; Zwinderman, Aeilko H.; Kastelein, John J. P.; Feskens, Edith J. M.; Bergen, Arthur A. B.
CORPORATE SOURCE: Department of Cardiology, University of Amsterdam, Amsterdam, Neth.
SOURCE: Circulation (2002), 106(7), 773-775
CODEN: CIRCAZ; ISSN: 0009-7322
PUBLISHER: Lippincott Williams & Wilkins
DOCUMENT TYPE: Journal
LANGUAGE: English
REFERENCE COUNT: 16 THERE ARE 16 CITED REFERENCES AVAILABLE FOR THIS RECORD. ALL CITATIONS AVAILABLE IN THE RE FORMAT

L3 ANSWER 24 OF 68 CAPLUS COPYRIGHT 2007 ACS on STN
ACCESSION NUMBER: 2001:776537 CAPLUS
DOCUMENT NUMBER: 136:292478
TITLE: A spectrum of ABCC6 mutations is responsible for pseudoxanthoma elasticum
AUTHOR(S): Le Saux, Olivier; Beck, Konstanze; Sachsinger,

Christine; Silvestri, Chiara; Treiber, Carina; Goring, Harald H. H.; Johnson, Eric W.; De Paepe, Anne; Pope, F. Michael; Pasquali-Ronchetti, Ivonne; Bercovitch, Lionel; Terry, Sharon; Boyd, Charles D.
CORPORATE SOURCE: Pacific Biomedical Research Center, University of Hawaii, Honolulu, HI, 96822, USA
SOURCE: American Journal of Human Genetics (2001), 69(4), 749-764
CODEN: AJHGAG; ISSN: 0002-9297
PUBLISHER: University of Chicago Press
DOCUMENT TYPE: Journal
LANGUAGE: English
REFERENCE COUNT: 64 THERE ARE 64 CITED REFERENCES AVAILABLE FOR THIS RECORD. ALL CITATIONS AVAILABLE IN THE RE FORMAT

L3 ANSWER 25 OF 68 CAPLUS COPYRIGHT 2007 ACS on STN
ACCESSION NUMBER: 2000:507713 CAPLUS
DOCUMENT NUMBER: 133:250501
TITLE: Homozygosity for the R1268Q Mutation in MRP6 , the pseudoxanthoma elasticum Gene, Is Not Disease-Causing
AUTHOR(S): Germain, Dominique P.; Perdu, Jerome; Remones, Veronique; Jeunemaitre, Xavier
CORPORATE SOURCE: Departement de Genetique, Hop. European Georges Pompidou, Universite Paris VI, Paris, Fr.
SOURCE: Biochemical and Biophysical Research Communications (2000), 274(2), 297-301
CODEN: BBRCA9; ISSN: 0006-291X
PUBLISHER: Academic Press
DOCUMENT TYPE: Journal
LANGUAGE: English
REFERENCE COUNT: 17 THERE ARE 17 CITED REFERENCES AVAILABLE FOR THIS RECORD. ALL CITATIONS AVAILABLE IN THE RE FORMAT

L3 ANSWER 26 OF 68 CAPLUS COPYRIGHT 2007 ACS on STN
ACCESSION NUMBER: 2000:397370 CAPLUS
DOCUMENT NUMBER: 133:118224
TITLE: Mutations in ABCC6 cause pseudoxanthoma elasticum
AUTHOR(S): Bergen, Arthur A. B.; Plomp, Astrid S.; Schuurman, Ellen J.; Terry, Sharon; Breuning, Martijn; Dauwerse, Hans; Swart, Jaap; Kool, Marcel; Van Soest, Simone; Baas, Frank; ten Brink, Jacoline B.; De Jong, Paulus T. V. M.
CORPORATE SOURCE: The Netherlands Ophthalmic Research Institute, Amsterdam, Neth.
SOURCE: Nature Genetics (2000), 25(2), 228-231
CODEN: NGENEC; ISSN: 1061-4036
PUBLISHER: Nature America Inc.
DOCUMENT TYPE: Journal
LANGUAGE: English
REFERENCE COUNT: 23 THERE ARE 23 CITED REFERENCES AVAILABLE FOR THIS RECORD. ALL CITATIONS AVAILABLE IN THE RE FORMAT

L3 ANSWER 27 OF 68 CAPLUS COPYRIGHT 2007 ACS on STN
ACCESSION NUMBER: 2000:397369 CAPLUS
DOCUMENT NUMBER: 133:118223
TITLE: Mutations in a gene encoding an ABC transporter cause pseudoxanthoma elasticum
AUTHOR(S): Le Saux, Olivier; Urban, Zsolt; Tschuch, Cordula; Csiszar, Katalin; Bacchelli, Barbara; Quaglino, Daniela; Pasquali-Ronchetti, Ivonne; Pope, F. Michael; Richards, Allan; Terry, Sharon; Bercovitch, Lionel; De Paepe, Anne; Boyd, Charles D.

CORPORATE SOURCE: Laboratory of Matrix Pathobiology, Pacific Biomedical Research Center, University of Hawai'i, Honolulu, HI, USA
SOURCE: Nature Genetics (2000), 25(2), 223-227
CODEN: NGENEC; ISSN: 1061-4036
PUBLISHER: Nature America Inc.
DOCUMENT TYPE: Journal
LANGUAGE: English
REFERENCE COUNT: 26 THERE ARE 26 CITED REFERENCES AVAILABLE FOR THIS RECORD. ALL CITATIONS AVAILABLE IN THE RE FORMAT

L3 ANSWER 28 OF 68 EMBASE COPYRIGHT (c) 2007 Elsevier B.V. All rights reserved on STN
ACCESSION NUMBER: 2006064091 EMBASE
TITLE: Role of serum fetuin-A, a major inhibitor of systemic calcification, in pseudoxanthoma elasticum.
AUTHOR: Hendig D.; Schulz V.; Arndt M.; Szliska C.; Kleesiek K.; Gotting C.
CORPORATE SOURCE: C. Gotting, Institut fur Laboratoriums- und Transfusionsmedizin, Herz- und Diabeteszentrum Nordrhein-Westfalen, Universitatsklinik der Ruhr-Universitat Bochum, Georgstrasse 11, 32545 Bad Oeynhausen, Germany. cgoetting@hdz-nrw.de
SOURCE: Clinical Chemistry, (2006) Vol. 52, No. 2, pp. 227-234. .
Refs: 53
ISSN: 0009-9147 CODEN: CLCHAU
COUNTRY: United States
DOCUMENT TYPE: Journal; Article
FILE SEGMENT: 005 General Pathology and Pathological Anatomy
013 Dermatology and Venereology
018 Cardiovascular Diseases and Cardiovascular Surgery
022 Human Genetics
LANGUAGE: English
SUMMARY LANGUAGE: English
ENTRY DATE: Entered STN: 31 Mar 2006
Last Updated on STN: 31 Mar 2006

L3 ANSWER 29 OF 68 EMBASE COPYRIGHT (c) 2007 Elsevier B.V. All rights reserved on STN
ACCESSION NUMBER: 2005556531 EMBASE
TITLE: Elevated xylosyltransferase I activities in pseudoxanthoma elasticum (PXE) patients as a marker of stimulated proteoglycan biosynthesis.
AUTHOR: Gotting C.; Hendig D.; Adam A.; Schon S.; Schulz V.; Szliska C.; Kuhn J.; Kleesiek K.
CORPORATE SOURCE: Dr. C. Gotting, Institut fur Laboratoriums-und Transfusionsmedizin, Herz-und Diabeteszentrum Nordrhein-Westfalen, Universitatsklinik der Ruhr-Universitat Bochum, Georgstrasse 11, 32545 Bad Oeynhausen, Germany. cgoetting@hdz-nrw.de
SOURCE: Journal of Molecular Medicine, (2005) Vol. 83, No. 12, pp. 984-992. .
Refs: 53
ISSN: 0946-2716 CODEN: JMLME8
COUNTRY: Germany
DOCUMENT TYPE: Journal; Article
FILE SEGMENT: 013 Dermatology and Venereology
022 Human Genetics
029 Clinical Biochemistry
LANGUAGE: English
SUMMARY LANGUAGE: English
ENTRY DATE: Entered STN: 12 Jan 2006
Last Updated on STN: 12 Jan 2006

L3 ANSWER 30 OF 68 EMBASE COPYRIGHT (c) 2007 Elsevier B.V. All rights reserved on STN

ACCESSION NUMBER: 2005481993 EMBASE
TITLE: Novel mutations in the ABCC6 gene of German patients with pseudoxanthoma elasticum.
AUTHOR: Schulz V.; Hendig D.; Szliska C.; Gotting C.; Kleesiek K.
CORPORATE SOURCE: V. Schulz, Institut fur Laboratoriums- und Transfusionsmedizin, Herz- und Diabeteszentrum Nordrhein-Westfalen, Universitatsklinik der Ruhr-Universitat Bochum, Georgstrasse 11, 32545 Bad Oeynhausen, Germany
SOURCE: Human Biology, (2005) Vol. 77, No. 3, pp. 367-384. .
Refs: 32
ISSN: 0018-7143 CODEN: HUBIAA
COUNTRY: United States
DOCUMENT TYPE: Journal; General Review
FILE SEGMENT: 012 Ophthalmology
013 Dermatology and Venereology
018 Cardiovascular Diseases and Cardiovascular Surgery
022 Human Genetics
LANGUAGE: English
SUMMARY LANGUAGE: English
ENTRY DATE: Entered STN: 28 Nov 2005
Last Updated on STN: 28 Nov 2005

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ACCESSION NUMBER: 2005168725 EMBASE
TITLE: Patients with premature coronary artery disease who carry the ABCC6 R1141X mutation have no Pseudoxanthoma Elasticum phenotype.
AUTHOR: Wegman J.J.; Hu X.; Tan H.; Bergen A.A.B.; Trip M.D.; Kastelein J.J.P.; Smulders Y.M.
CORPORATE SOURCE: Y.M. Smulders, Department of Internal Medicine, VU University Medical Center, De Boelelaan 1117, 1081 HV, Amsterdam, Netherlands. Y.Smulders@VUMC.NL
SOURCE: International Journal of Cardiology, (28 Apr 2005) Vol. 100, No. 3, pp. 389-393. .
Refs: 25
ISSN: 0167-5273 CODEN: IJCDD5
COUNTRY: Ireland
DOCUMENT TYPE: Journal; Article
FILE SEGMENT: 005 General Pathology and Pathological Anatomy
018 Cardiovascular Diseases and Cardiovascular Surgery
022 Human Genetics
LANGUAGE: English
SUMMARY LANGUAGE: English
ENTRY DATE: Entered STN: 19 May 2005
Last Updated on STN: 19 May 2005

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ACCESSION NUMBER: 2004518942 EMBASE
TITLE: Efficient molecular diagnostic strategy for ABCC6 in pseudoxanthoma elasticum.
AUTHOR: Hu X.; Plomp A.; Gorgels T.; Ten Brink J.; Loves W.; Mannens M.; De Jong P.T.V.M.; Bergen A.A.B.
CORPORATE SOURCE: Dr. A.A.B. Bergen, The Netherlands Ophthal. Res. Inst., Department of Ophthalmogenetics, Meibergdreef 47, 1105 BA Amsterdam, Netherlands. a.bergen@ioi.knaw.nl
SOURCE: Genetic Testing, (2004) Vol. 8, No. 3, pp. 292-300. .
Refs: 24
ISSN: 1090-6576 CODEN: GETEF4
COUNTRY: United States

DOCUMENT TYPE: Journal; Article
FILE SEGMENT: 005 General Pathology and Pathological Anatomy
022 Human Genetics
031 Arthritis and Rheumatism
LANGUAGE: English
SUMMARY LANGUAGE: English
ENTRY DATE: Entered STN: 28 Dec 2004
Last Updated on STN: 28 Dec 2004

L3 ANSWER 33 OF 68 EMBASE COPYRIGHT (c) 2007 Elsevier B.V. All rights reserved on STN
ACCESSION NUMBER: 2004374944 EMBASE
TITLE: [From gene to disease; pseudoxanthoma elasticum and the ABCC6 gene].
VAN GEN NAAR ZIEKTE; PSEUDOXANTHOMA ELASTICUM EN HET ABCC6-GEN.
AUTHOR: Bergen A.A.B.; Plomp A.S.; Gorgels T.G.M.F.; De Jong P.T.V.M.
CORPORATE SOURCE: Dr. A.A.B. Bergen, Interuniv. Oogheelkundig Instituut, Meibergdreef 47, 1105 BA Amsterdam, Netherlands. a.bergen@ioi.knaw.nl
SOURCE: Nederlands Tijdschrift voor Geneeskunde, (7 Aug 2004) Vol. 148, No. 32, pp. 1586-1589. .
Refs: 3
ISSN: 0028-2162 CODEN: NETJAN
COUNTRY: Netherlands
DOCUMENT TYPE: Journal; Article
FILE SEGMENT: 022 Human Genetics
LANGUAGE: Dutch
SUMMARY LANGUAGE: English
ENTRY DATE: Entered STN: 24 Sep 2004
Last Updated on STN: 24 Sep 2004

L3 ANSWER 34 OF 68 EMBASE COPYRIGHT (c) 2007 Elsevier B.V. All rights reserved on STN
ACCESSION NUMBER: 2004164516 EMBASE
TITLE: Novel ABCC6 mutations in pseudoxanthoma elasticum.
AUTHOR: Chassaing N.; Martin L.; Mazereeuw J.; Barrie L.; Nizard S.; Bonafe J.-L.; Calvas P.; Hovnanian A.
CORPORATE SOURCE: A. Hovnanian, Department of Medical Genetics, Pavilion Lefebvre, Purpan Hospital, Place du Dr Baylac, 31059 Toulouse Cedex 09, France. alain.hovnanian@toulouse.inserm.fr
SOURCE: Journal of Investigative Dermatology, (2004) Vol. 122, No. 3, pp. 608-613. .
Refs: 26
ISSN: 0022-202X CODEN: JIDEAE
COUNTRY: United States
DOCUMENT TYPE: Journal; Article
FILE SEGMENT: 013 Dermatology and Venereology
022 Human Genetics
LANGUAGE: English
SUMMARY LANGUAGE: English
ENTRY DATE: Entered STN: 13 May 2004
Last Updated on STN: 13 May 2004

L3 ANSWER 35 OF 68 EMBASE COPYRIGHT (c) 2007 Elsevier B.V. All rights reserved on STN
ACCESSION NUMBER: 2003506646 EMBASE
TITLE: Multidrug resistance protein-6 (MRP6) in human dermal fibroblasts. Comparison between cells from normal subjects and from Pseudoxanthoma elasticum patients.

AUTHOR: Boraldi F.; Quaglino D.; Croce M.A.; Garcia Fernandez M.I.; Tiozzo R.; Gheduzzi D.; Bacchelli B.; Pasquali Ronchetti I.
CORPORATE SOURCE: I. Pasquali Ronchetti, Department of Biomedical Sciences, Univ. of Modena and Reggio Emilia, via Campi, 287, 41100 Modena, Italy. ronchetti.ivonne@unimore.it
SOURCE: Matrix Biology, (2003) Vol. 22, No. 6, pp. 491-500. .
Refs: 35
ISSN: 0945-053X CODEN: MTBOEC
COUNTRY: Netherlands
DOCUMENT TYPE: Journal; Article
FILE SEGMENT: 030 Pharmacology
037 Drug Literature Index
LANGUAGE: English
SUMMARY LANGUAGE: English
ENTRY DATE: Entered STN: 16 Jan 2004
Last Updated on STN: 16 Jan 2004

L3 ANSWER 36 OF 68 EMBASE
ACCESSION NUMBER: 2003176365 EMBASE
TITLE: Analysis of the frequent R1141X mutation in the ABCC6 gene in pseudoxanthoma elasticum.
AUTHOR: Hu X.; Peek R.; Plomp A.; Ten Brink J.; Scheffer G.; Van Soest S.; Leys A.; De Jong P.T.V.M.; Bergen A.A.B.
CORPORATE SOURCE: A.A.B. Bergen, Department of Ophthalmogenetics, Netherlands Ophthal. Res. Institute, Meibergdreef 47, 1105 BA Amsterdam, Netherlands. a.bergen@ioi.knaw.nl
SOURCE: Investigative Ophthalmology and Visual Science, (1 May 2003) Vol. 44, No. 5, pp. 1824-1829. .
Refs: 24
ISSN: 0146-0404 CODEN: IOVSDA
COUNTRY: United States
DOCUMENT TYPE: Journal; Article
FILE SEGMENT: 012 Ophthalmology
013 Dermatology and Venereology
022 Human Genetics
LANGUAGE: English
SUMMARY LANGUAGE: English
ENTRY DATE: Entered STN: 22 May 2003
Last Updated on STN: 22 May 2003

L3 ANSWER 37 OF 68 EMBASE
ACCESSION NUMBER: 2003172049 EMBASE
TITLE: ABCC6/MRP6 mutations: Further insight into the molecular pathology of pseudoxanthoma elasticum.
AUTHOR: Hu X.; Plomp A.; Wijnholds J.; ten Brink J.; van Soest S.; van den Born L.I.; Leys A.; Peek R.; de Jong P.T.V.M.; Bergen A.A.B.
CORPORATE SOURCE: Dr. A.A.B. Bergen, Netherlands Ophthalmic Res. Inst., Department of Ophthalmogenetics, Meibergdreef 47, 1105 BA Amsterdam, Netherlands. a.bergen@ioi.knaw.nl
SOURCE: European Journal of Human Genetics, (1 Mar 2003) Vol. 11, No. 3, pp. 215-224. .
Refs: 41
ISSN: 1018-4813 CODEN: EJHGEU
COUNTRY: United Kingdom
DOCUMENT TYPE: Journal; Article
FILE SEGMENT: 005 General Pathology and Pathological Anatomy
022 Human Genetics
031 Arthritis and Rheumatism
LANGUAGE: English
SUMMARY LANGUAGE: English

ENTRY DATE: Entered STN: 19 May 2003
Last Updated on STN: 19 May 2003

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ACCESSION NUMBER: 2002359883 EMBASE
TITLE: Frequent mutation in the ABCC6 gene (R1141X) is associated with a strong increase in the prevalence of coronary artery disease.
AUTHOR: Trip M.D.; Smulders Y.M.; Wegman J.J.; Hu X.; Boer J.M.A.; Ten Brink J.B.; Zwinderman A.H.; Kastelein J.J.P.; Feskens E.J.M.; Bergen A.A.B.
CORPORATE SOURCE: Dr. M.D. Trip, Department of Cardiology, Academic Medical Centre, Meibergdreef 9, 1105 AZ Amsterdam, Netherlands. M.D.Trip@AMC.UVA.NL
SOURCE: Circulation, (13 Aug 2002) Vol. 106, No. 7, pp. 773-775. .
Refs: 16
ISSN: 0009-7322 CODEN: CIRCAZ
COUNTRY: United States
DOCUMENT TYPE: Journal; Article
FILE SEGMENT: 018 Cardiovascular Diseases and Cardiovascular Surgery
022 Human Genetics
LANGUAGE: English
SUMMARY LANGUAGE: English
ENTRY DATE: Entered STN: 24 Oct 2002
Last Updated on STN: 24 Oct 2002

L3 ANSWER 39 OF 68 EMBASE COPYRIGHT (c) 2007 Elsevier B.V. All rights reserved on STN
ACCESSION NUMBER: 2001329372 EMBASE
TITLE: A spectrum of ABCC6 mutations is responsible for pseudoxanthoma elasticum.
AUTHOR: Saux O.L.; Beck K.; Sachsinger C.; Silvestri C.; Treiber C.; Goring H.H.H.; Johnson E.W.; De Paepe A.; Pope F.M.; Pasquali-Ronchetti I.; Bercovitch L.; Terry S.; Boyd C.D.
CORPORATE SOURCE: Dr. C.D. Boyd, Laboratory of Matrix Pathobiology, Pacific Biomedical Research Center, University of Hawai'i, 1993 East-West Road, Honolulu, HI 96822, United States. cbkc08901@aol.com
SOURCE: American Journal of Human Genetics, (2001) Vol. 69, No. 4, pp. 749-764. .
Refs: 64
ISSN: 0002-9297 CODEN: AJHGAG
COUNTRY: United States
DOCUMENT TYPE: Journal; Article
FILE SEGMENT: 013 Dermatology and Venereology
021 Developmental Biology and Teratology
022 Human Genetics
029 Clinical Biochemistry
LANGUAGE: English
SUMMARY LANGUAGE: English
ENTRY DATE: Entered STN: 11 Oct 2001
Last Updated on STN: 11 Oct 2001

L3 ANSWER 40 OF 68 EMBASE COPYRIGHT (c) 2007 Elsevier B.V. All rights reserved on STN
ACCESSION NUMBER: 2000277899 EMBASE
TITLE: Homozygosity for the R1268Q mutation in MRP6, the pseudoxanthoma elasticum gene, is not disease-causing.
AUTHOR: Germain D.P.; Perdu J.; Remones V.; Jeunemaitre X.
CORPORATE SOURCE: D.P. Germain, Laboratoire de Genetique, Hopital Broussais, 96 rue Didot, 75014 Paris, France. dominique.germain@brs.ap-hop-paris.fr

SOURCE: Biochemical and Biophysical Research Communications, (2 Aug 2000) Vol. 274, No. 2, pp. 297-301. .
Refs: 17
ISSN: 0006-291X CODEN: BBRCA
COUNTRY: United States
DOCUMENT TYPE: Journal; Article
FILE SEGMENT: 005 General Pathology and Pathological Anatomy
013 Dermatology and Venereology
022 Human Genetics
LANGUAGE: English
SUMMARY LANGUAGE: English
ENTRY DATE: Entered STN: 24 Aug 2000
Last Updated on STN: 24 Aug 2000

L3 ANSWER 41 OF 68 BIOTECHDS COPYRIGHT 2007 THE THOMSON CORP. on STN
ACCESSION NUMBER: 2002-11434 BIOTECHDS
TITLE: Frequent mutation in the ABCC6 gene (R1141X) is associated with a strong increase in the prevalence of coronary artery disease;
ATP-binding cassette transporter gene expression profiling, useful for useful Pseudoxanthoma elasticum therapy and diagnosis
AUTHOR: TRIP MD; SMULDERS YM; WEGMAN JJ; HU XF; BOER JMA; TEN BRINK JB; ZWINDERMAN AH; KASTELEIN JJP; FESKENS EJM; BERGEN AAB
CORPORATE SOURCE: Univ Amsterdam; Univ Amsterdam; Univ Amsterdam; Univ Amsterdam; Natl Inst Publ Hlth and Environm; Netherlands Ophthalm Res Inst
LOCATION: Trip MD, Univ Amsterdam, Acad Med Ctr, Dept Cardiol, Meibergdreef 9, NL-1105 AZ Amsterdam, Netherlands
SOURCE: CIRCULATION; (2002) 106, 7, 773-775
ISSN: 0009-7322
DOCUMENT TYPE: Journal
LANGUAGE: English

L3 ANSWER 42 OF 68 SCISEARCH COPYRIGHT (c) 2007 The Thomson Corporation on STN
ACCESSION NUMBER: 2006:147134 SCISEARCH
THE GENUINE ARTICLE: 008VU
TITLE: Role of serum fetuin-A, a major inhibitor of systemic calcification, in pseudoxanthoma elasticum
AUTHOR: Hendig D; Schulz V; Arndt M; Szliska C; Kleesiek K; Gotting C (Reprint)
CORPORATE SOURCE: Ruhr Univ Bochum, Inst Lab & Transfus Med Herz, Diabet Zentrum Nordrhein Westfalen, Univ Klin, Georgstr 11, D-32545 Bad Oeynhausen, Germany (Reprint); Ruhr Univ Bochum, Inst Lab & Transfus Med Herz, Diabet Zentrum Nordrhein Westfalen, Univ Klin, D-32545 Bad Oeynhausen, Germany; Krankenhaus Bethesda, Dermatol Klin, Freudenberg, Germany
cgoetting@hdz-nrw.de
COUNTRY OF AUTHOR: Germany
SOURCE: CLINICAL CHEMISTRY, (FEB 2006) Vol. 52, No. 2, pp. 227-234
ISSN: 0009-9147.
PUBLISHER: AMER ASSOC CLINICAL CHEMISTRY, 2101 L STREET NW, SUITE 202, WASHINGTON, DC 20037-1526 USA.
DOCUMENT TYPE: Article; Journal
LANGUAGE: English
REFERENCE COUNT: 53
ENTRY DATE: Entered STN: 16 Feb 2006
Last Updated on STN: 16 Feb 2006
ABSTRACT IS AVAILABLE IN THE ALL AND IALL FORMATS

L3 ANSWER 43 OF 68 SCISEARCH COPYRIGHT (c) 2007 The Thomson Corporation on

STN
ACCESSION NUMBER: 2006:52157 SCISEARCH
THE GENUINE ARTICLE: 998UN
TITLE: Elevated xylosyltransferase I activities in pseudoxanthoma elasticum (PXE) patients as a marker of stimulated proteoglycam biosynthesis
AUTHOR: Gotting C (Reprint); Hendig D; Adam A; Schon S; Schulz V; Szliska C; Kuhn J; Kleesiek K
CORPORATE SOURCE: Ruhr Univ Bochum, Inst Lab & Transfus Med, Herz & Diabet Zentrum Nordrhein Westfalen, Univ Klin, Georgstr 11, D-32545 Bad Oeynhausen, Germany (Reprint); Ruhr Univ Bochum, Inst Lab & Transfus Med, Herz & Diabet Zentrum Nordrhein Westfalen, Univ Klin, D-32545 Bad Oeynhausen, Germany; Krankenhaus Bethesda, Dermatol Klin, Freudenberg, Germany
cgoetting@hdz-nrw.de
COUNTRY OF AUTHOR: Germany
SOURCE: JOURNAL OF MOLECULAR MEDICINE-JMM, (DEC 2005) Vol. 83, No. 12, pp. 984-992.
ISSN: 0946-2716.
PUBLISHER: SPRINGER, 233 SPRING STREET, NEW YORK, NY 10013 USA.
DOCUMENT TYPE: Article; Journal
LANGUAGE: English
REFERENCE COUNT: 53
ENTRY DATE: Entered STN: 19 Jan 2006
Last Updated on STN: 19 Jan 2006
ABSTRACT IS AVAILABLE IN THE ALL AND IALL FORMATS

L3 ANSWER 44 OF 68 SCISEARCH COPYRIGHT (c) 2007 The Thomson Corporation on STN
ACCESSION NUMBER: 2005:1132818 SCISEARCH
THE GENUINE ARTICLE: 982TO
TITLE: Novel mutations in the ABCC6 gene of German patients with Pseudoxanthoma elasticum
AUTHOR: Schulz V (Reprint); Hendig D; Szliska C; Gotting C; Kleesiek K
CORPORATE SOURCE: Ruhr Univ Bochum, Univ Klin, Inst Lab & Transfusionsmed Herz & Diabeteszentrum, Georgstr 11, D-32545 Bad Oeynhausen, Germany (Reprint); Ruhr Univ Bochum, Univ Klin, Inst Lab & Transfusionsmed Herz & Diabeteszentrum, D-32545 Bad Oeynhausen, Germany; Krankenhaus Bethesda, Dermatol Klin, Freudenberg, Germany
COUNTRY OF AUTHOR: Germany
SOURCE: HUMAN BIOLOGY, (JUN 2005) Vol. 77, No. 3, pp. 367-384.
ISSN: 0018-7143.
PUBLISHER: WAYNE STATE UNIV PRESS, 4809 WOODWARD AVE, DETROIT, MI 48201-1309 USA.
DOCUMENT TYPE: Article; Journal
LANGUAGE: English
REFERENCE COUNT: 32
ENTRY DATE: Entered STN: 24 Nov 2005
Last Updated on STN: 24 Nov 2005
ABSTRACT IS AVAILABLE IN THE ALL AND IALL FORMATS

L3 ANSWER 45 OF 68 SCISEARCH COPYRIGHT (c) 2007 The Thomson Corporation on STN
ACCESSION NUMBER: 2005:486490 SCISEARCH
THE GENUINE ARTICLE: 922CX
TITLE: Patients with premature coronary artery disease who carry the ABCC6 R1141X mutation have no Pseudoxanthoma Elasticum phenotype
AUTHOR: Wegman J J; Hu X F; Tan H R; Bergen A A B; Trip M D; Kastelein J J P; Smulders Y M (Reprint)
CORPORATE SOURCE: Vrije Univ Amsterdam, Ctr Med, Dept Internal Med, De

Boelelaan 1117, NL-1081 HV Amsterdam, Netherlands (Reprint); Vrije Univ Amsterdam, Ctr Med, Dept Internal Med, NL-1081 HV Amsterdam, Netherlands; Univ Amsterdam, Acad Med Ctr, Dept Vasc Med, NL-1105 AZ Amsterdam, Netherlands; Netherlands Ophthalm Res Inst, NL-1100 AC Amsterdam, Netherlands; Univ Amsterdam, Acad Med Ctr, Dept Ophthalmol, NL-1105 AZ Amsterdam, Netherlands; Univ Amsterdam, Acad Med Ctr, Dept Cardiol, NL-1105 AZ Amsterdam, Netherlands
Y.Smulders@VUMC.NL

COUNTRY OF AUTHOR: Netherlands

SOURCE: INTERNATIONAL JOURNAL OF CARDIOLOGY, (28 APR 2005) Vol. 100, No. 3, pp. 389-393.
ISSN: 0167-5273.

PUBLISHER: ELSEVIER IRELAND LTD, ELSEVIER HOUSE, BROOKVALE PLAZA, EAST PARK SHANNON, CO, CLARE, 00000, IRELAND.

DOCUMENT TYPE: Article; Journal

LANGUAGE: English

REFERENCE COUNT: 25

ENTRY DATE: Entered STN: 22 May 2005
Last Updated on STN: 22 May 2005

ABSTRACT IS AVAILABLE IN THE ALL AND IALL FORMATS

L3 ANSWER 46 OF 68 SCISEARCH COPYRIGHT (c) 2007 The Thomson Corporation on STN

ACCESSION NUMBER: 2004:1058494 SCISEARCH

THE GENUINE ARTICLE: 875QR

TITLE: Efficient molecular diagnostic strategy for ABCC6 in pseudoxanthoma elasticum

AUTHOR: Hu X F; Plomp A; Gorgels T; Ten Brink J; Loves W; Mannens M; De Jong P T V M; Bergen A A B (Reprint)

CORPORATE SOURCE: Netherlands Ophthalm Res Inst, Dept Ophthalmogenet, KNAW, Meibergdreef 47, NL-1105 BA Amsterdam, Netherlands (Reprint); Netherlands Ophthalm Res Inst, Dept Ophthalmogenet, KNAW, NL-1105 BA Amsterdam, Netherlands; AMC, Dept Clin Genet, Amsterdam, Netherlands; AMC, Dept Ophthalmol, Amsterdam, Netherlands; EUR, Dept Epidemiol & Biostat, Rotterdam, Netherlands
a.bergen@ioi.knaw.nl

COUNTRY OF AUTHOR: Netherlands

SOURCE: GENETIC TESTING, (FAL 2004) Vol. 8, No. 3, pp. 292-300.
ISSN: 1090-6576.

PUBLISHER: MARY ANN LIEBERT INC, 2 MADISON AVENUE, LARCHMONT, NY 10538 USA.

DOCUMENT TYPE: Article; Journal

LANGUAGE: English

REFERENCE COUNT: 24

ENTRY DATE: Entered STN: 30 Dec 2004
Last Updated on STN: 30 Dec 2004

ABSTRACT IS AVAILABLE IN THE ALL AND IALL FORMATS

L3 ANSWER 47 OF 68 SCISEARCH COPYRIGHT (c) 2007 The Thomson Corporation on STN

ACCESSION NUMBER: 2004:640671 SCISEARCH

THE GENUINE ARTICLE: 809UB

TITLE: Novel ABCC6 mutations in pseudoxanthoma elasticum

AUTHOR: Chassaing N; Martin L; Mazereeuw J; Barrie L; Nizard S; Bonafe J L; Calvas P; Hovnanian A (Reprint)

CORPORATE SOURCE: Hop Purpan, Dept Med Genet, Pavill Lefebvre, Pl Dr Baylac, F-31059 Toulouse 09, France (Reprint); Hop Purpan, Dept Med Genet, Pavill Lefebvre, F-31059 Toulouse 09, France; Hop Purpan, INSERM, Pavill Lefebvre, U563, F-31059 Toulouse, France; Porte Madeleine Hosp, Dept Dermatol,

Orleans, France; Hop Rangueil, Dept Dermatol, Toulouse, France; Porte Madeleine Hosp, Dept Med Genet, Orleans, France
hovnanian@toulouse.inserm.fr
COUNTRY OF AUTHOR: France
SOURCE: JOURNAL OF INVESTIGATIVE DERMATOLOGY, (MAR 2004) Vol. 122, No. 3, pp. 608-613.
ISSN: 0022-202X.
PUBLISHER: BLACKWELL PUBLISHING INC, 350 MAIN ST, MALDEN, MA 02148 USA.
DOCUMENT TYPE: Article; Journal
LANGUAGE: English
REFERENCE COUNT: 26
ENTRY DATE: Entered STN: 6 Aug 2004
Last Updated on STN: 6 Aug 2004
ABSTRACT IS AVAILABLE IN THE ALL AND IALL FORMATS

L3 ANSWER 48 OF 68 SCISEARCH COPYRIGHT (c) 2007 The Thomson Corporation on STN
ACCESSION NUMBER: 2004:409583 SCISEARCH
THE GENUINE ARTICLE: 816IH
TITLE: Does autosomal dominant pseudoxanthoma elasticum exist?
AUTHOR: Plomp A S; Hu X F; de Jong P T V M (Reprint); Bergen A A B
CORPORATE SOURCE: Netherlands Ophthalm Res Inst, KNAW, Meibergdreef 47, NL-1105 BA Amsterdam, Netherlands (Reprint); Netherlands Ophthalm Res Inst, KNAW, NL-1105 BA Amsterdam, Netherlands; Univ Amsterdam, Acad Med Ctr, Dept Clin Genet, NL-1105 AZ Amsterdam, Netherlands; Univ Amsterdam, Acad Med Ctr, Dept Ophthalmol, NL-1105 AZ Amsterdam, Netherlands; Erasmus Med Ctr Rotterdam, Inst Epidemiol & Biostat, Rotterdam, Netherlands
COUNTRY OF AUTHOR: Netherlands
SOURCE: AMERICAN JOURNAL OF MEDICAL GENETICS PART A, (1 MAY 2004) Vol. 126A, No. 4, pp. 403-412.
ISSN: 0148-7299.
PUBLISHER: WILEY-LISS, DIV JOHN WILEY & SONS INC, 605 THIRD AVE, NEW YORK, NY 10158-0012 USA.
DOCUMENT TYPE: Article; Journal
LANGUAGE: English
REFERENCE COUNT: 44
ENTRY DATE: Entered STN: 21 May 2004
Last Updated on STN: 21 May 2004
ABSTRACT IS AVAILABLE IN THE ALL AND IALL FORMATS

L3 ANSWER 49 OF 68 SCISEARCH COPYRIGHT (c) 2007 The Thomson Corporation on STN
ACCESSION NUMBER: 2004:41234 SCISEARCH
THE GENUINE ARTICLE: 756FX
TITLE: Multidrug resistance protein-6 (MRP6) in human dermal fibroblasts. Comparison between cells from normal subjects and from Pseudoxanthoma elasticum patients
AUTHOR: Boraldi F; Quaglino D; Croce M A; Fernandez M I G; Tiozzo R; Gheduzzi D; Bacchelli B; Ronchetti I P (Reprint)
CORPORATE SOURCE: Univ Modena, Dept Biomed Sci, Via Campi 287, I-41100 Modena, Italy (Reprint); Univ Modena, Dept Biomed Sci, I-41100 Modena, Italy
COUNTRY OF AUTHOR: Italy
SOURCE: MATRIX BIOLOGY, (NOV 2003) Vol. 22, No. 6, pp. 491-500.
ISSN: 0945-053X.
PUBLISHER: ELSEVIER SCIENCE BV, PO BOX 211, 1000 AE AMSTERDAM, NETHERLANDS.
DOCUMENT TYPE: Article; Journal

LANGUAGE: English
REFERENCE COUNT: 35
ENTRY DATE: Entered STN: 16 Jan 2004
Last Updated on STN: 16 Jan 2004
ABSTRACT IS AVAILABLE IN THE ALL AND IALL FORMATS

L3 ANSWER 50 OF 68 SCISEARCH COPYRIGHT (c) 2007 The Thomson Corporation on STN
ACCESSION NUMBER: 2003:376640 SCISEARCH
THE GENUINE ARTICLE: 672CQ
TITLE: Analysis of the frequent R1141X mutation in the ABCC6 gene in pseudoxanthoma elasticum
AUTHOR: Hu X F; Peek R; Plomp A; ten Brink J; Scheffer G; van Soest S; Leys A; de Jong P T V M; Bergen A A B (Reprint)
CORPORATE SOURCE: Royal Netherlands Acad Art & Sci, Netherlands Ophthalm Res Inst, Dept Ophthalmogenet, Meibergdreef 47, NL-1105 BA Amsterdam, Netherlands (Reprint); Royal Netherlands Acad Art & Sci, Netherlands Ophthalm Res Inst, Dept Ophthalmogenet, NL-1105 BA Amsterdam, Netherlands; Univ Amsterdam, Acad Med Ctr, Dept Clin Genet, NL-1105 AZ Amsterdam, Netherlands; Free Univ Amsterdam, Dept Pathol, Amsterdam, Netherlands; Katholieke Univ Leuven, Dept Ophthalmol, Louvain, Belgium; Univ Amsterdam, Dept Ophthalmol, Amsterdam, Netherlands; Erasmus Med Ctr, Inst Epidemiol & Biostat, Rotterdam, Netherlands
COUNTRY OF AUTHOR: Netherlands; Belgium
SOURCE: INVESTIGATIVE OPHTHALMOLOGY & VISUAL SCIENCE, (MAY 2003) Vol. 44, No. 5, pp. 1824-1829.
ISSN: 0146-0404.
PUBLISHER: ASSOC RESEARCH VISION OPHTHALMOLOGY INC, 12300 TWINBROOK PARKWAY, ROCKVILLE, MD 20852-1606 USA.
DOCUMENT TYPE: Article; Journal
LANGUAGE: English
REFERENCE COUNT: 24
ENTRY DATE: Entered STN: 16 May 2003
Last Updated on STN: 16 May 2003
ABSTRACT IS AVAILABLE IN THE ALL AND IALL FORMATS

L3 ANSWER 51 OF 68 SCISEARCH COPYRIGHT (c) 2007 The Thomson Corporation on STN
ACCESSION NUMBER: 2002:704526 SCISEARCH
THE GENUINE ARTICLE: 585NK
TITLE: Frequent mutation in the ABCC6 gene (R1141X) is associated with a strong increase in the prevalence of coronary artery disease
AUTHOR: Trip M D (Reprint); Smulders Y M; Wegman J J; Hu X F; Boer J M A; ten Brink J B; Zwinderman A H; Kastelein J J P; Feskens E J M; Bergen A A B
CORPORATE SOURCE: Univ Amsterdam, Acad Med Ctr, Dept Cardiol, Meibergdreef 9, NL-1105 AZ Amsterdam, Netherlands (Reprint); Univ Amsterdam, Acad Med Ctr, Dept Cardiol, NL-1105 AZ Amsterdam, Netherlands; Univ Amsterdam, Acad Med Ctr, Dept Clin Epidemiol & Biostat, NL-1105 AZ Amsterdam, Netherlands; Univ Amsterdam, Acad Med Ctr, Dept Vasc Med, NL-1105 AZ Amsterdam, Netherlands; Univ Amsterdam, Acad Med Ctr, Dept Clin Genet, NL-1105 AZ Amsterdam, Netherlands; Natl Inst Publ Hlth & Environm, Dept Chron Dis Epidemiol, NL-3720 BA Bilthoven, Netherlands; Netherlands Ophthalm Res Inst, NL-1100 AC Amsterdam, Netherlands
COUNTRY OF AUTHOR: Netherlands
SOURCE: CIRCULATION, (13 AUG 2002) Vol. 106, No. 7, pp. 773-775.
ISSN: 0009-7322.
PUBLISHER: LIPPINCOTT WILLIAMS & WILKINS, 530 WALNUT ST,

PHILADELPHIA, PA 19106-3621 USA.
DOCUMENT TYPE: Article; Journal
LANGUAGE: English
REFERENCE COUNT: 16
ENTRY DATE: Entered STN: 13 Sep 2002
Last Updated on STN: 13 Sep 2002
ABSTRACT IS AVAILABLE IN THE ALL AND IALL FORMATS

L3 ANSWER 52 OF 68 SCISEARCH COPYRIGHT (c) 2007 The Thomson Corporation on STN
ACCESSION NUMBER: 2001:757747 SCISEARCH
THE GENUINE ARTICLE: 472WP
TITLE: A spectrum of ABCC6 mutations is responsible for pseudoxanthoma elasticum
AUTHOR: Le Saux O; Beck K; Sachsinger C; Silvestri C; Treiber C; Goring H H H; Johnson E W; De Paepe A; Pope F M; Pasquali-Ronchetti I; Bercovitch L; Terry S; Boyd C D (Reprint)
CORPORATE SOURCE: Univ Hawaii, Pacific Biomed Res Ctr, Lab Matrix Pathobiol, 1993 East West Rd, Honolulu, HI 96822 USA (Reprint); Univ Hawaii, Pacific Biomed Res Ctr, Lab Matrix Pathobiol, Honolulu, HI 96822 USA; Univ Modena, Dept Biomed Sci, I-41100 Modena, Italy; SW Fdn Biomed Res, Dept Genet, San Antonio, TX USA; Barrow Neurol Inst, Phoenix, AZ 85013 USA; State Univ Ghent Hosp, Ctr Med Genet, B-9000 Ghent, Belgium; Univ Wales Hosp, Inst Med Genet, MRC, Connect Tissue Genet Grp, Cardiff CF4 4XN, S Glam, Wales; Brown Med Sch, Dept Dermatol, Providence, RI USA; PXE Int Inc, Sharon, MA USA
COUNTRY OF AUTHOR: USA; Italy; Belgium; Wales
SOURCE: AMERICAN JOURNAL OF HUMAN GENETICS, (OCT 2001) Vol. 69, No. 4, pp. 749-764.
ISSN: 0002-9297.
PUBLISHER: UNIV CHICAGO PRESS, 1427 E 60TH ST, CHICAGO, IL 60637-2954 USA.
DOCUMENT TYPE: Article; Journal
LANGUAGE: English
REFERENCE COUNT: 64
ENTRY DATE: Entered STN: 5 Oct 2001
Last Updated on STN: 5 Oct 2001
ABSTRACT IS AVAILABLE IN THE ALL AND IALL FORMATS

L3 ANSWER 53 OF 68 SCISEARCH COPYRIGHT (c) 2007 The Thomson Corporation on STN
ACCESSION NUMBER: 2000:608985 SCISEARCH
THE GENUINE ARTICLE: 341LU
TITLE: Homozygosity for the R1268Q mutation in MRP6, the pseudoxanthoma elasticum gene, is not disease-causing
AUTHOR: Germain D P (Reprint); Perdu J; Remones V; Jeunemaitre X
CORPORATE SOURCE: Hop Broussais, Lab Genet, 96 Rue Didot, F-75014 Paris, France (Reprint); Univ Paris 06, Hop Europeen Georges Pompidou, Dept Genet, Paris, France
COUNTRY OF AUTHOR: France
SOURCE: BIOCHEMICAL AND BIOPHYSICAL RESEARCH COMMUNICATIONS, (2 AUG 2000) Vol. 274, No. 2, pp. 297-301.
ISSN: 0006-291X.
PUBLISHER: ACADEMIC PRESS INC, 525 B ST, STE 1900, SAN DIEGO, CA 92101-4495 USA.
DOCUMENT TYPE: Article; Journal
LANGUAGE: English
REFERENCE COUNT: 17
ENTRY DATE: Entered STN: 2000
Last Updated on STN: 2000

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L3 ANSWER 54 OF 68 BIOSIS COPYRIGHT (c) 2007 The Thomson Corporation on STN
ACCESSION NUMBER: 2006:264773 BIOSIS
DOCUMENT NUMBER: PREV200600264108
TITLE: Role of serum fetuin-A, a major inhibitor of systemic calcification, in pseudoxanthoma elasticum.
AUTHOR(S): Hendig, Doris; Schulz, Veronika; Arndt, Marius; Szliska, Christiane; Kleesiek, Knut; Goetting, ChriStian [Reprint Author]
CORPORATE SOURCE: Ruhr Univ Bochum, Inst Lab and Transfus Med Herz, Diabet Zentrum Nordrhein Westfalen, Univ Klin, Georgstr 11, D-32545 Bad Oeynhausen, Germany
cgoetting@hdz-nrw.de
SOURCE: Clinical Chemistry, (FEB 2006) Vol. 52, No. 2, pp. 227-234.
CODEN: CLCHAU. ISSN: 0009-9147.
DOCUMENT TYPE: Article
LANGUAGE: English
ENTRY DATE: Entered STN: 10 May 2006
Last Updated on STN: 10 May 2006

L3 ANSWER 55 OF 68 BIOSIS COPYRIGHT (c) 2007 The Thomson Corporation on STN
ACCESSION NUMBER: 2006:179130 BIOSIS
DOCUMENT NUMBER: PREV200600168444
TITLE: Elevated xylosyltransferase I activities in pseudoxanthoma elasticum (PXE) patients as a marker of stimulated proteoglycam biosynthesis.
AUTHOR(S): Goetting, Christian [Reprint Author]; Hendig, Doris; Adam, Alexandra; Schoen, Sylvia; Schulz, Veronika; Szliska, Christiane; Kuhn, Joachim; Kleesiek, Knut
CORPORATE SOURCE: Ruhr Univ Bochum, Inst Lab and Transfus Med, Herz and Diabet Zentrum Nordrhein Westfalen, Univ Klin, Georgstr 11, D-32545 Bad Oeynhausen, Germany
cgoetting@hdz-nrw.de
SOURCE: Journal of Molecular Medicine (Berlin), (DEC 2005) Vol. 83, No. 12, pp. 984-992.
ISSN: 0946-2716. E-ISSN: 1432-1440.
DOCUMENT TYPE: Article
LANGUAGE: English
ENTRY DATE: Entered STN: 9 Mar 2006
Last Updated on STN: 9 Mar 2006

L3 ANSWER 56 OF 68 BIOSIS COPYRIGHT (c) 2007 The Thomson Corporation on STN
ACCESSION NUMBER: 2006:86400 BIOSIS
DOCUMENT NUMBER: PREV200600087300
TITLE: Novel mutations in the ABCC6 gene of German patients with Pseudoxanthoma elasticum.
AUTHOR(S): Schulz, Veronika [Reprint Author]; Hendig, Doris; Szliska, Christiane; Goetting, Christian; Kleesiek, Knut
CORPORATE SOURCE: Ruhr Univ Bochum, Univ Klin, Inst Lab and Transfusionsmed Herz and Diabeteszentrum, Georgstr 11, D-32545 Bad Oeynhausen, Germany
SOURCE: Human Biology, (JUN 2005) Vol. 77, No. 3, pp. 367-384.
CODEN: HUBIAA. ISSN: 0018-7143.
DOCUMENT TYPE: Article
LANGUAGE: English
ENTRY DATE: Entered STN: 25 Jan 2006
Last Updated on STN: 25 Jan 2006

L3 ANSWER 57 OF 68 BIOSIS COPYRIGHT (c) 2007 The Thomson Corporation on STN

ACCESSION NUMBER: 2005:319112 BIOSIS
DOCUMENT NUMBER: PREV200510114507
TITLE: Genotype-phenotype correlation in 62 patients with pseudoxanthoma elasticum.
AUTHOR(S): Fuchsel, L. [Reprint Author]; Kozic, H.; McGuigan, K.; Skvarka, C.; Jacobson, M.; Uitto, J.; Ringpfeil, F.
CORPORATE SOURCE: Jefferson Med Coll, Philadelphia, PA USA
SOURCE: Journal of Investigative Dermatology, (MAR 2004) Vol. 122, No. 3, pp. A93.
Meeting Info.: 65th Annual Meeting of the Society-for-Investigative-Dermatology. Providence, RI, USA. April 28 -May 01, 2004. Soc Investigat Dermatol.
CODEN: JIDEAE. ISSN: 0022-202X.
DOCUMENT TYPE: Conference; (Meeting)
Conference; Abstract; (Meeting Abstract)
LANGUAGE: English
ENTRY DATE: Entered STN: 25 Aug 2005
Last Updated on STN: 25 Aug 2005

L3 ANSWER 58 OF 68 BIOSIS COPYRIGHT (c) 2007 The Thomson Corporation on STN
ACCESSION NUMBER: 2005:319073 BIOSIS
DOCUMENT NUMBER: PREV200510114468
TITLE: dHPLC screening detects novel and recurrent mutations in pseudoxanthoma elasticum.
AUTHOR(S): Fratta, S. [Reprint Author]; Ringpfeil, F.; Terry, S.; Terry, P.; Uitto, J.; Pfendner, E. G.
CORPORATE SOURCE: Thomas Jefferson Univ, Philadelphia, PA 19107 USA
SOURCE: Journal of Investigative Dermatology, (MAR 2004) Vol. 122, No. 3, pp. A87,A86.
Meeting Info.: 65th Annual Meeting of the Society-for-Investigative-Dermatology. Providence, RI, USA. April 28 -May 01, 2004. Soc Investigat Dermatol.
CODEN: JIDEAE. ISSN: 0022-202X.
DOCUMENT TYPE: Conference; (Meeting)
Conference; Abstract; (Meeting Abstract)
LANGUAGE: English
ENTRY DATE: Entered STN: 25 Aug 2005
Last Updated on STN: 25 Aug 2005

L3 ANSWER 59 OF 68 BIOSIS COPYRIGHT (c) 2007 The Thomson Corporation on STN
ACCESSION NUMBER: 2005:261978 BIOSIS
DOCUMENT NUMBER: PREV200510046658
TITLE: Patients with premature coronary artery disease who carry the ABCC6 R1141X mutation have no Pseudoxanthoma Elasticum phenotype.
AUTHOR(S): Wegman, Jurgen J.; Hu, Xiaofeng; Tan, Hendra; Bergen, Arthur A. B.; Trip, Mieke D.; Kastelein, John J. P.; Smulders, Yvo M. [Reprint Author]
CORPORATE SOURCE: Vrije Univ Amsterdam, Ctr Med, Dept Internal Med, De Boelelaan 1117, NL-1081 HV Amsterdam, Netherlands
Y.Smulders@VUMC.NL
SOURCE: International Journal of Cardiology, (APR 28 2005) Vol. 100, No. 3, pp. 389-393.
CODEN: IJCDD5. ISSN: 0167-5273.
DOCUMENT TYPE: Article
LANGUAGE: English
ENTRY DATE: Entered STN: 14 Jul 2005
Last Updated on STN: 14 Jul 2005

L3 ANSWER 60 OF 68 BIOSIS COPYRIGHT (c) 2007 The Thomson Corporation on STN
ACCESSION NUMBER: 2005:67464 BIOSIS

DOCUMENT NUMBER: PREV200500068254
TITLE: Efficient molecular diagnostic strategy for ABCC6 in pseudoxanthoma elasticum.
AUTHOR(S): Hu, Xiaofeng; Plomp, Astrid; Gorgels, Theo; ten Brink, Jacoline; Loves, Willem; Mannens, Marcel; de Jong, Paulus T. V. M.; Bergen, Arthur A. B. [Reprint Author]
CORPORATE SOURCE: Dept OphthalmogenetKNAW, Netherlands Ophthalm Res Inst, Meibergdreef 47, NL-1105 BA, Amsterdam, Netherlands a.bergen@ioi.knaw.nl
SOURCE: Genetic Testing, (Autumn 2004) Vol. 8, No. 3, pp. 292-300. print.
ISSN: 1090-6576 (ISSN print).
DOCUMENT TYPE: Article
LANGUAGE: English
ENTRY DATE: Entered STN: 9 Feb 2005
Last Updated on STN: 9 Feb 2005

L3 ANSWER 61 OF 68 BIOSIS COPYRIGHT (c) 2007 The Thomson Corporation on STN
ACCESSION NUMBER: 2004:396192 BIOSIS
DOCUMENT NUMBER: PREV200400396457
TITLE: Novel ABCC6 mutations in pseudoxanthoma elasticum.
AUTHOR(S): Chassaing, Nicolas; Martin, Ludovic; Mazereeuw, Juliette; Barri, Laurence; Nizard, Sonia; Bonafe, Jean-Louis; Calvas, Patrick; Hovnanian, Alain [Reprint Author]
CORPORATE SOURCE: Dept Med GenetPavill Lefebvre, Hop Purpan, Pl Dr Baylac, F-31059, Toulouse, 09, France hovnanian@toulouse.inserm.fr
SOURCE: Journal of Investigative Dermatology, (March 2004) Vol. 122, No. 3, pp. 608-613. print.
ISSN: 0022-202X (ISSN print).
DOCUMENT TYPE: Article
LANGUAGE: English
ENTRY DATE: Entered STN: 13 Oct 2004
Last Updated on STN: 13 Oct 2004

L3 ANSWER 62 OF 68 BIOSIS COPYRIGHT (c) 2007 The Thomson Corporation on STN
ACCESSION NUMBER: 2004:91139 BIOSIS
DOCUMENT NUMBER: PREV200400092355
TITLE: Multidrug resistance protein-6 (MRP6) in human dermal fibroblasts. Comparison between cells from normal subjects and from Pseudoxanthoma elasticum patients.
AUTHOR(S): Boraldi, F.; Quaglino, D.; Croce, M. A.; Fernandez, M. I. Garcia; Tiozzo, R.; Gheduzzi, D.; Bacchelli, B.; Ronchetti, I. Pasquali [Reprint Author]
CORPORATE SOURCE: Department of Biomedical Sciences, University of Modena and Reggio Emilia, via Campi, 287, 41100, Modena, Italy ronchetti.ivonne@unimore.it
SOURCE: Matrix Biology, (November 2003) Vol. 22, No. 6, pp. 491-500. print.
ISSN: 0945-053X (ISSN print).
DOCUMENT TYPE: Article
LANGUAGE: English
ENTRY DATE: Entered STN: 11 Feb 2004
Last Updated on STN: 11 Feb 2004

L3 ANSWER 63 OF 68 BIOSIS COPYRIGHT (c) 2007 The Thomson Corporation on STN
ACCESSION NUMBER: 2003:262254 BIOSIS
DOCUMENT NUMBER: PREV200300262254
TITLE: ABCC6/MRP6 mutations: Further insight

into the molecular pathology of pseudoxanthoma elasticum.
AUTHOR(S): Hu, Xiaofeng; Plomp, Astrid; Wijnholds, Jan; ten Brink, Jacoline; van Soest, Simone; van den Born, L. Ingeborgh; Leys, Anita; Peek, Ron; de Jong, Paulus T. V. M.; Bergen, Arthur A. B. [Reprint Author]
CORPORATE SOURCE: Department of Ophthalmogenetics, The Netherlands Ophthalmic Research Institute, Meibergdreef 47, 1105 BA, Amsterdam, Netherlands
a.bergen@ioi.knaw.nl
SOURCE: European Journal of Human Genetics, (March 2003) Vol. 11, No. 3, pp. 215-224. print.
ISSN: 1018-4813.
DOCUMENT TYPE: Article
LANGUAGE: English
ENTRY DATE: Entered STN: 4 Jun 2003
Last Updated on STN: 4 Jun 2003

L3 ANSWER 64 OF 68 BIOSIS COPYRIGHT (c) 2007 The Thomson Corporation on STN
ACCESSION NUMBER: 2003:256347 BIOSIS
DOCUMENT NUMBER: PREV200300256347
TITLE: Analysis of the frequent R1141X mutation in the ABCC6 gene in pseudoxanthoma elasticum.
AUTHOR(S): Hu, Xiaofeng; Peek, Ron; Plomp, Astrid; ten Brink, Jacoline; Scheffer, George; van Soest, Simone; Leys, Anita; de Jong, Paulus T. V. M.; Bergen, Arthur A. B. [Reprint Author]
CORPORATE SOURCE: Department of Ophthalmogenetics, Netherlands Ophthalmic Research Institute, Meibergdreef 47, 1105 BA, Amsterdam, Netherlands
a.bergen@ioi.knaw.nl
SOURCE: IOVS, (May 2003) Vol. 44, No. 5, pp. 1824-1829. print.
DOCUMENT TYPE: Article
LANGUAGE: English
OTHER SOURCE: GenBank-U91318
ENTRY DATE: Entered STN: 28 May 2003
Last Updated on STN: 28 May 2003

L3 ANSWER 65 OF 68 BIOSIS COPYRIGHT (c) 2007 The Thomson Corporation on STN
ACCESSION NUMBER: 2003:165025 BIOSIS
DOCUMENT NUMBER: PREV200300165025
TITLE: Molecular analysis of Pseudoxanthoma Elasticum: spectrum of ABCC6 gene mutations in the Netherlands.
AUTHOR(S): Hu, X. [Reprint Author]; Plomp, A. [Reprint Author]; Ten Brink, J. B. [Reprint Author]; Wijnholds, J. [Reprint Author]; Schuurman, E. J. [Reprint Author]; Soest, S. van [Reprint Author]; Oud, M. [Reprint Author]; Peek, R. [Reprint Author]; Jong, P. T. V. M. [Reprint Author]; Bergen, A. A. B. [Reprint Author]
CORPORATE SOURCE: Research Unit Ophthalmogenetics, Netherlands Ophthalmic Research Institute, Amsterdam, Netherlands
SOURCE: ARVO Annual Meeting Abstract Search and Program Planner, (2002) Vol. 2002, pp. Abstract No. 2394. cd-rom.
Meeting Info.: Annual Meeting of the Association For Research in Vision and Ophthalmology. Fort Lauderdale, Florida, USA. May 05-10, 2002.
DOCUMENT TYPE: Conference; (Meeting)
Conference; Abstract; (Meeting Abstract)
LANGUAGE: English
ENTRY DATE: Entered STN: 2 Apr 2003
Last Updated on STN: 2 Apr 2003

L3 ANSWER 66 OF 68 BIOSIS COPYRIGHT (c) 2007 The Thomson Corporation on STN

ACCESSION NUMBER: 2002:471764 BIOSIS
DOCUMENT NUMBER: PREV200200471764
TITLE: Frequent mutation in the ABCC6 gene (R1141X) is associated with a strong increase in the prevalence of coronary artery disease.
AUTHOR(S): Trip, Mieke D. [Reprint author]; Smulders, Yvo M.; Wegman, Jurgen J.; Hu, Xiaofeng; Boer, Jolanda M. A.; ten Brink, Jacoline B.; Zwinderman, Aeilko H.; Kastelein, John J. P.; Feskens, Edith J. M.; Bergen, Arthur A. B.
CORPORATE SOURCE: Department of Cardiology, Academic Medical Centre, Meibergdreef 9, 1105 AZ, Amsterdam, Netherlands M.D.Trip@AMC.UVA.NL
SOURCE: Circulation, (August 13, 2002) Vol. 106, No. 7, pp. 773-775. print.
CODEN: CIRCAZ. ISSN: 0009-7322.
DOCUMENT TYPE: Article
LANGUAGE: English
ENTRY DATE: Entered STN: 11 Sep 2002
Last Updated on STN: 11 Sep 2002

L3 ANSWER 67 OF 68 BIOSIS COPYRIGHT (c) 2007 The Thomson Corporation on STN

ACCESSION NUMBER: 2001:482882 BIOSIS
DOCUMENT NUMBER: PREV200100482882
TITLE: A spectrum of ABCC6 mutations is responsible for pseudoxanthoma elasticum.
AUTHOR(S): Le Saux, Olivier; Beck, Konstanze; Sachsinger, Christine; Silvestri, Chiara; Treiber, Carina; Goring, Harald H. H.; Johnson, Eric W.; De Paepe, Anne; Pope, F. Michael; Pasquali-Ronchetti, Ivonne; Bercovitch, Lionel; Terry, Sharon; Boyd, Charles D. [Reprint author]
CORPORATE SOURCE: Laboratory of Matrix Pathobiology, Pacific Biomedical Research Center, University of Hawai'i, 1993 East-West Road, Honolulu, HI, 96822, USA cbkc08901@aol.com
SOURCE: American Journal of Human Genetics, (October, 2001) Vol. 69, No. 4, pp. 749-764. print.
CODEN: AJHGAG. ISSN: 0002-9297.
DOCUMENT TYPE: Article
LANGUAGE: English
OTHER SOURCE: Genbank-AC002039; Genbank-AC002045; Genbank-AC002492; Genbank-NM000352; Genbank-NM000972; Genbank-NM001171; Genbank-NM00492; Genbank-NT010393; Genbank-U91318; Genbank-XM017599; Genbank-XM017612
ENTRY DATE: Entered STN: 17 Oct 2001
Last Updated on STN: 25 Feb 2002

L3 ANSWER 68 OF 68 BIOSIS COPYRIGHT (c) 2007 The Thomson Corporation on STN

ACCESSION NUMBER: 2000:410284 BIOSIS
DOCUMENT NUMBER: PREV200000410284
TITLE: Homozygosity for the R1268Q mutation in MRP6, the pseudoxanthoma elasticum gene, is not disease-causing.
AUTHOR(S): Germain, Dominique P. [Reprint author]; Perdu, Jerome; Remones, Veronique; Jeunemaitre, Xavier
CORPORATE SOURCE: Laboratoire de Genetique, Hopital Broussais, 96, Rue Didot, 75014, Paris, France
SOURCE: Biochemical and Biophysical Research Communications, (August 2, 2000) Vol. 274, No. 2, pp. 297-301. print.
CODEN: BBRCA9. ISSN: 0006-291X.

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☐	L1	6780587.pn.	2
☐	L2	PXE mutation or (pseudoxanthoma near elasticum)	484
☐	L3	10/764328	1
☐	L4	ABCC6 allele	3
☐	L5	ABCC6	38
☐	L6	L5 and MRP6	14
☐	L7	L6 and L2	6
☐	L8	R1141X or 3421C>T	30828236
☐	L9	L8 and L2	484
☐	L10	L9 and MRP6	6

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DOCUMENT TYPE: Article
LANGUAGE: English
ENTRY DATE: Entered STN: 27 Sep 2000
Last Updated on STN: 8 Jan 2002

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